

A Dissertation on

**“A STUDY OF NUCHAL TRANSLUCENCY IN  
EARLY PREGNANCY”**



*Dissertation submitted to*

**THE TAMILNADU Dr.M.G.R. MEDICAL UNIVERSITY**

**CHENNAI – 600 032**

*With partial fulfillment of the regulations*

*For the award of the degree of*

**M. S. (OBSTETRICS AND GYNAECOLOGY)**



**COIMBATORE MEDICAL COLLEGE**

**COIMBATORE**

**APRIL 2015**

*CERTIFICATE*

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This is to certify that the dissertation entitled **“A STUDY OF NUCHAL TRANSLUCENCY IN EARLY PREGNANCY”** is a bonafide work done by Dr. PADMA DEVI R, in partial fulfillment of the requirement for the MS Degree in the Department of Obstetrics and Gynaecology, Coimbatore Medical College Hospital, Coimbatore.

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## **DECLARATION**

I **Dr.Padma Devi.R**, solemnly declare that the dissertation entitled “**A STUDY OF NUCHAL TRANSLUCENCY IN EARLY PREGNANCY**” was done by me in the Department of Obstetrics and Gynaecology at Coimbatore Medical College Hospital during the period from August 2013 to July 2014 under the guidance & supervision of **Dr.R. Manonmani, M.D., D.G.O.**, Associate Professor, Department of Obstetrics and Gynaecology, Coimbatore Medical College Hospital, Coimbatore. The dissertation is submitted to The Tamil nadu Dr.MGR Medical University, Chennai towards the partial fulfillment of the requirement for the award of M.S., degree in Obstetrics and Gynaecology. I have not submitted this dissertation on any previous occasion to any university for the award of any degree.

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## *ACKNOWLEDGEMENT*

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## ACKNOWLEDGEMENT

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# A STUDY ON NUCHAL TRANSLUCENCY IN EARLY PREGNANCY

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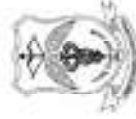
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## *LIST OF ABBREVIATIONS*

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## LIST OF ABBREVIATIONS USED

CVS	→	Chorion Villus Sampling
NT	→	Nuchal Translucency
PAPP-A	→	Pregnancy Associated Plasma Protein - A
$\beta$ HCG	→	$\beta$ Human Chorionic Gonadotrophin
UE3	→	Unconjugated Estriol
MSAFP	→	Maternal Serum $\alpha$ Feto protein
IUD	→	Intrauterine Death
PROM	→	Premature Rupture of Membranes
PPROM	→	Preterm Premature Rupture of Membranes
GDM	→	Gestational Diabetes Mellitus
GHT	→	Gestational Hypertension
IUGR	→	Intrauterine Growth Restriction
MAS	→	Meconium Aspiration Syndrome
LSCS	→	Lower Segment Caesarean Section
MA	→	Maternal Age
NB	→	Nasal Bone
ASD	→	Atrial Septal Defect
LMP	→	Last Menstrual Period

EDD	→	Expected Date of Delivery
CHD	→	Congenital Heart Disease
TCV	→	Tricuspid Valve
DV	→	Ductus Venosus
FMF	→	Fronto Maxillary Facial angle



# *ABSTRACT*

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## **ABSTRACT**

### **BACKGROUND**

Screening for fetal abnormalities is necessary for early, timely identification to take appropriate intervention and management to achieve optimal perinatal outcome.

Screening of fetal nuchal translucency at 10-14 weeks of gestation identifies the fetuses at risk for chromosomal abnormalities, cardiac defects, structural defects, genetic syndromes.

### **OBJECTIVES**

- To detect whether nuchal translucency can be used as early screening tool to identify fetal abnormalities.
- To detect increased Nuchal translucency in women with singleton pregnancy between 10-14 weeks of gestation and its usefulness in identifying the fetus at risk for chromosomal abnormalities and other defects.

### **METHODS**

We at Coimbatore Medical College Hospital have undertaken a prospective clinical study in 50 pregnant women.

Association of an increased nuchal translucency with fetal abnormalities were evaluated in our study.

## **RESULTS**

On statistical analysis, our study showed that there is association between increased nuchal translucency and fetal abnormalities.

## **INTERPRETATION AND CONCLUSION**

This study has given us an insight to use of fetal nuchal translucency as a screening tool to identify fetuses at risk for chromosomal abnormalities, cardiac defects, and structural defects at 10-14 weeks of gestation. Thus pushing prenatal diagnosis of fetal abnormalities to I trimester rather than II trimester, which gives the affected women an option of early termination, which is more acceptable and emotionally less traumatic to the expectant mothers.

## **KEYWORDS**

Nuchal Translucency; Chorion Villus Sampling; Cordocentesis; Amniocentesis; Down's syndrome; Aneuploidy; Fetal Abnormalities; Cardiac defects.

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# *INTRODUCTION*

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## INTRODUCTION

One of the important cause of perinatal death and childhood handicap is chromosomal abnormalities.<sup>1</sup> The incidence of significant chromosomal abnormalities and birth defect is 3% , out of which 66% accounts to Down's syndrome<sup>2</sup>, 25% of severe mental retardation in children is due to Down's syndrome. The frequency throughout the world is about 0-13% of births. The incidence in India is 1 in 600-700. Down's babies survives with mental and physical disabilities and causes mental trauma to family. Down's syndrome screening becomes essential and it is done prenatally to reduce the live birth of Down's babies.<sup>3</sup>

Prenatal screening has been increasing over the past 20 years for Down's syndrome and other aneuploidies with amniocentesis, chorionic villus sampling, cordocentesis. Previously screening was done among women of advanced maternal age (age > 35 years) and those with previously affected pregnancies. Second trimester Biochemical and sonographic markers and soft markers for Down's syndrome were offered to women at increased risk for Down's syndrome.<sup>4</sup> This combined approach yields sensitivities for screening to 67-76% for a false positive

rate of 5%.<sup>5</sup> Chromosomal abnormalities contribute to 50% of first trimester miscarriages.<sup>8</sup>

The choice of definitive diagnosis of aneuploidy is limited to amniocentesis which is performed at 15 weeks of gestation at the earliest and prenatal diagnosis is made in the second trimester. Furthermore, amniocentesis do not detect 25% of Down's syndrome and 5% false positive rate ensures that for every single case of Down's syndrome to be detected, 60 amniocentesis procedures were performed. The pregnancy loss rate is 1 in 200 with amniocentesis and about 1 normal fetus is lost for every 3 fetuses with Down's syndrome detected.<sup>6</sup>

First trimester screening includes Nuchal Translucency measurement, presence of nasal bone and fronto maxillary facial angle measurement Sonographically, and biochemical markers like PAPP-A and  $\beta$ -HCG, and Doppler study of tricuspid valve and ductus venosus.<sup>7</sup> Also fetal anatomy can be systematically examined and major structural anomalies can be detected at this stage and also helps in accurate pregnancy dating. With these screening facilities, prenatal screening has moved from second trimester to first trimester screening.

Thickened Nuchal fold in Down's syndrome fetus was demonstrated by Beryl Benacerraf in 1985 after which sonographic screening for aneuploidy has become evident.<sup>9</sup>

It was Dr. Langdon Down, 100 years back has reported too large swelling at the back of neck in affected fetuses. Sonographically this swelling at the back of neck is studied as Nuchal Translucency (Down's syndrome was named after his name). NT measurement is best done at 10-14 weeks of gestational age.<sup>10</sup> In addition to Down's syndrome, other trisomies, other chromosomal defects, major cardiac defects, skeletal defects and various genetic syndromes are associated with increased Nuchal translucency.<sup>11</sup>

## *AIMS AND OBJECTIVES*

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## **AIMS AND OBJECTIVES OF STUDY**

### **Aim:**

To detect whether nuchal translucency can be used as an early screening tool to identify fetal abnormalities.

### **Objective:**

To detect increased Nuchal translucency in women with singleton pregnancy between 10-14 weeks of gestation and its usefulness in identifying fetus at risk for chromosomal abnormalities and other defects.



# *REVIEW OF LITERATURE*

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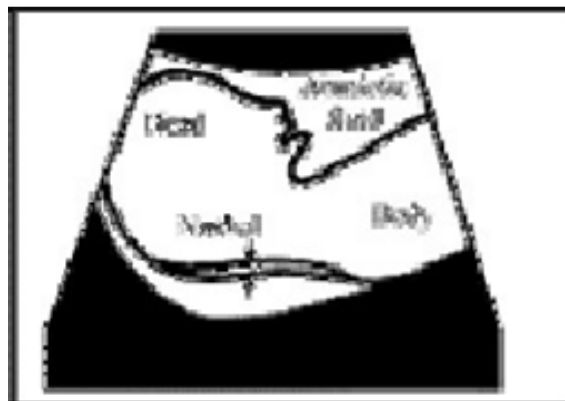
## REVIEW OF LITERATURE

### **Nuchal Translucency:**

It is the maximum thickness of subcutaneous translucency measured between the skin and soft tissue overlying the cervical spine of fetus.

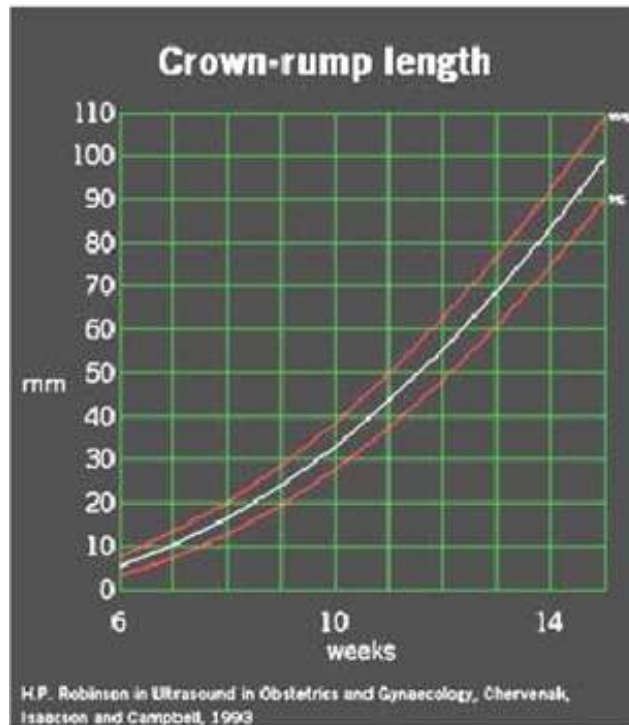


**Fig. 1: I Trimester Fetus 1**

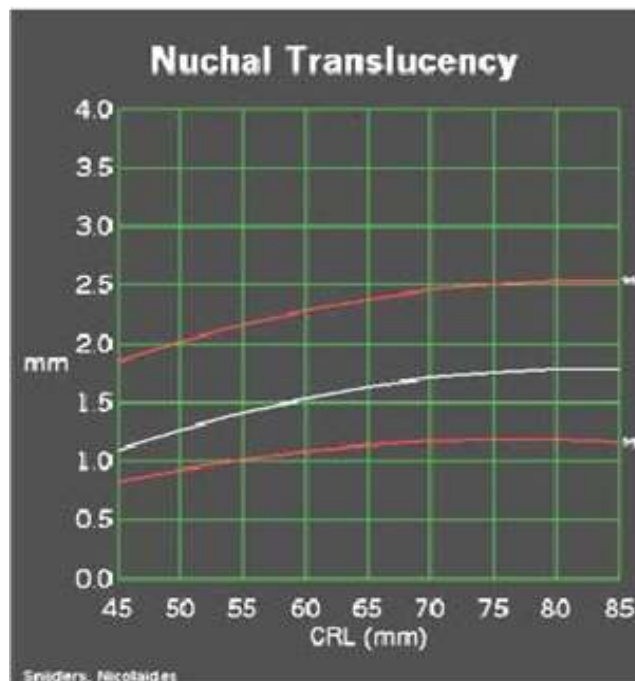


**Fig 2. Diagrammatic representation of the fetal Nuchal translucency**



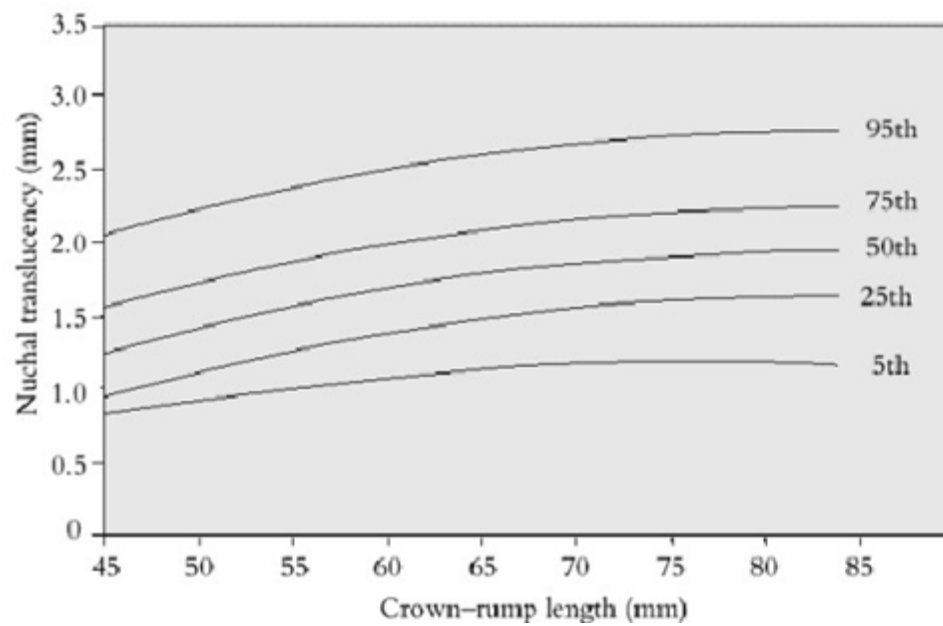


**Fig. 4: CRL vs. Gestational age (weeks)**



**Fig. 5: Nuchal Translucency normally increases with gestation (CRL)**

Nuchal translucency rises with increasing gestational age. The cut off on this progressive rise is used as 95<sup>th</sup> centile corresponding to gestational age, which results in more sensitive and specific indicator for detection of anomalous fetus.<sup>15, 16</sup>



**Fig. 6: NT vs. CRL**

In 1992, Nicolaides KH, has been credited for his improvements in this field.<sup>17</sup>



**Kypros H. Nicolaides**

KH Nicolades et al, in 1994 conducted a study among 1273 pregnant women with singleton pregnancy. First trimester ultrasound was done at 10-14 weeks of gestation. Association with fetal aneuploidy was noted with Nuchal translucency greater than or equal to 3mm with a sensitivity of 85%, specificity of 95.9% and positive predictive value of 35.5% with a false positive rate of 5%.<sup>18</sup>

Pandya et al, in 1995 conducted a study among 1763 pregnant women. First trimester ultrasound was done at 10-13 weeks of gestation. Association with fetal aneuploidy was noted with Nuchal translucency greater than or equal to 2.5mm with a

sensitivity of 75%, specificity of 92% and a false positive rate of 8%.<sup>19</sup>

Bewlert et al in 1995 conducted a study among 1127 pregnant women. First trimester ultrasound was done at 8-13 weeks of gestation. Association with fetal aneuploidy was noted with Nuchal translucency greater than or equal to 3mm with a sensitivity of 40%, specificity of 94% and a false positive rate of 6.1%.<sup>20</sup>

Taipele et al, in 1997 conducted a study among 10,010 pregnant women. First trimester ultrasound was done at 10-14 weeks of gestation. Association with fetal aneuploidy was noted with Nuchal translucency greater or equal to 3mm with a sensitivity of 62%, specificity of 99% and positive predictive value of 24% and negative predictive value of 100%.<sup>21</sup>

Economides et al, in 1998 conducted a study among 2281 pregnant women. First trimester ultrasound was done at 11-14 weeks of gestation. Association of fetal aneuploidy was noted with Nuchal translucency greater or equal to the 99<sup>th</sup> centile for gestational age with a sensitivity of 81%, specificity of 99% and a false positive rate of 0.4%.<sup>22</sup>

Snijders et al, in 1998 conducted a study among 127 pregnant women for assessment of risk with increasing maternal age. Study included women of median age of 31 years with singleton pregnancies and fetal nuchal translucency measurement was done at 10-14 weeks of gestation and reported a sensitivity of 77% and a false positive rate of 4.4%.<sup>23</sup>

Hafner et al, in 1998 conducted a study among 4233 pregnant women. First trimester ultrasound was done at 10-14 weeks of gestation. Association of fetal aneuploidy was noted with Nuchal translucency greater than 2.5mm with a sensitivity of 65%, specificity of 98.5% , positive predictive value of 14.8% and a negative predictive value of 99.9% and a false positive rate of 1.5%.<sup>24</sup>

Schwarzler et al, in 1999 conducted a study among 4523 pregnant women. First trimester screening for fetal aneuploidy and congenital heart defects by maternal age and nuchal translucency measurement at 10-14 weeks of gestation in ultrasound was done in unselected population, with a false positive rate of 4.7%, sensitivity of 76% in detecting fetal aneuploidy.<sup>25</sup>



## **INCREASED NUCHAL TRANSLUCENCY AND ITS ASSOCIATIONS:**

### **I. Chromosomal Defects:**

The prevalence of chromosomal defects increases exponentially with increasing Nuchal translucency thickness. Among these chromosomal defects, Trisomy 21 (i.e., Down 's syndrome) accounts for 50%, Trisomy 18 or 13 for 25%, Turners syndrome for 10%, Triploidy for 5%, other chromosomal defects for 10%.<sup>23</sup>

### **II. Fetal Abnormalities:**

There is association between increased NT and fetal abnormalities like aneuploidy , cardiac defects, skeletal defects and other defects. In chromosomally normal fetuses, with NT measurement <95<sup>th</sup> percentile, the prevalence of fetal abnormalities is 1.6%. It increases to 25% for NT between the 95<sup>th</sup> and 99<sup>th</sup> percentile and increases exponentially thereafter to about 45% for NT of 6.5mm or more.<sup>26,27</sup>

### **III. Fetal Death:**

The prevalence of fetal death increases exponentially with increasing nuchal translucency thickness in chromosomally normal fetuses. The prevalence is 1.3% in those with nuchal translucency between 95<sup>th</sup> and 99<sup>th</sup> centile and increases to about 20% for nuchal translucency of 6.5mm or more. Death occurs by 20 weeks of gestation in majority of fetuses and they even progress to severe hydrops.<sup>26, 27</sup>

### **IV. Developmental Delay:**

The prevalence of developmental delay is 2.4% in chromosomally and anatomically normal fetuses with increased nuchal translucency.

### **V. Chromosomally normal fetuses with increased NT:**

**A. Cardiac defects:** In both chromosomally normal and abnormal fetuses increased NT was associated with cardiac defects .(Hyett et al 1997,1999) In chromosomally normal fetuses, the prevalence of major cardiac defects increases exponentially with NT thickness from 16/1000

in those with NT below the 95th centile, to about 1% for NT of 2.5-3.4 mm, 3% for NT of 3.5-4.4mm, 7% for NT of 4.5-5.4mm, 20% for NT of 5.5-6.4 mm and 30% for NT of 6.5mm or more (Souka et al 2004, Hyett et al 1997, 1999).<sup>28,29,30</sup>

**B. Pulmonary defects:** Diaphragmatic hernia,

Cystic adenomatoid malformation

(Sebire et al 1997)<sup>31</sup>

**C. Abdominal wall Defect:** Cloacal exstrophy

Exomphalos

Gastroschisis (Snijders et al 1995)<sup>32</sup>

**D. Central Nervous System defects:**

- Acrania / anencephaly
- Agenesis of the corpus callosum
- Craniosynostosis
- Dandy walker malformation
- Encephalocele
- Fowler syndrome

- Holoprosencephaly
- Hydrolethalus syndrome
- Iniencephaly
- Joubert syndrome
- Macrocephaly
- Microcephaly
- Spina bifida
- Trigenocephaly C
- Ventriculomegaly

**E. Neuromuscular defects:**

- Myotonic dystrophy
- Spinal muscular atrophy

**F. Body stalk anomaly: (Daskalakis et al 1997)<sup>23</sup>**

**G. Gastrointestinal tract defects:**

- Crohn's disease
- Duodenal atresia
- Esophageal atresia
- Small bowel obstruction

**H. Genitourinary defects:**

- Ambiguous genitalia
- Congenital adrenal hyperplasia

- Congenital nephrotic syndrome
- Hydronephrosis
- Hypospadias
- Infantile polycystic kidneys
- Meckel-Gruber syndrome
- Megacystis
- Multicystic dysplastic kidneys
- Renal agenesis

#### **I. Skeletal defects:**

- Achondrogenesis
- Achondroplasia
- Asphyxiating thoracic dystrophy
- Blomstrand osteochondrodysplasia
- Campomelic dwarfism
- Cleidocranial dysplasia
- Hypochondroplasia
- Kyphoscoliosis
- Limb reduction defect
- Osteogenesis imperfecta
- Short-rib polydactyly syndrome
- Talipes equino varus

- Thanatophoric dwarfism
- VACTER association

**J. Metabolic defects:**

- Beckwith-Wiedemann syndrome
- GM1gangliosidosis
- Long-chain 3-hydroxyacyl-coenzyme A dehydrogenase deficiency
- Mucopolysaccharidosis type VII
- Smith-Lemli-Opitz syndrome
- Vitamin D resistant rickets
- Zellweger syndrome

**K. Facial defect:**

- Agnathia/ micrognathia
- Facial cleft
- Microphthalmia
- Treacher-Collins syndrome

**L. Nuchal defect:**

- Cystic hygroma
- Neck lipoma

**M. Cardiac defect:**

- Di George syndrome

**N. Fetal anemia:**

- Fetal anemia
- Blackfan Diamond anaemia
- Congenital erythropoietic porphyria
- Fanconi anemia
- Parvovirus B 19 infection
- Thalassaemia- $\alpha$

**VI. Genetic Syndromes<sup>34</sup>**

- Achondrogenesis
- Achondroplasia\*
- Adrenal hyperplasia\*
- Asphyxiating thoracic dystrophy
- Beckwith-Wiedemann syndrome
- Blackfan-Diamond anemia
- Blomstrand Osteochondrodysplasia
- Brachmann-Corneliade Lange syndrome
- Campomelic dysplasia
- CHARGE association
- Cleidocranial dysplasia

- Dyserythropoietic
- Erythropoietic Porphyria (Gunther's disease)
- Fanconi anemia
- Fowler syndrome
- Fryn syndrome
- GM 1-Gangliosidosis\*
- Hydroletharus syndrome
- Hypochondroplasia
- Infantile polycystic kidney disease
- Joubert syndrome
- Lymphedema
- Mucopolysaccharidosis type VII
- Myotonic dystrophy
- Nephritic syndrome
- Noonan syndrome
- Osteogenesis imperfecta type 11
- Perlman syndrome
- Roberts syndrome
- Smith-Lemli-opitz syndrome
- Spinal muscular atrophy type 1
- Stickler syndrome



- Thalassaemia- $\alpha$
- Thanatophoric dysplasia
- Treacher Collins syndrome
- Trigonocephaly 'C' syndrome
- VACTER association
- Vitamin D resistant rickets
- Zellweger syndrome

## **PATHOPHYSIOLOGY OF NUCHAL TRANSLUCENCY**

The various conditions associated with increased NT suggests that there may be several mechanisms for collection of fluid in the skin behind the cervical spine and the pathophysiology is not fully understood.

### **Proposed Theory:**

Transient cerebral hyperperfusion with associated diffusion of fluid across a primitive atlanto-occipital membrane in the first trimester contributes to NT and this might explain the transient nature.<sup>35</sup>

### **Other Theories:**

#### **1. Cardiac dysfunction**

There is high association between increased NT and abnormalities of heart and great arteries in both chromosomally abnormal and normal fetuses, suggesting that heart failure contributes to increased NT.

In fetuses, fluid collection behind the neck occurs much like it does in dependent ankle edema in later life. This occurs partly because of fetal tendency to lie on its back and partly because of the skin of the neck.<sup>36-39</sup>

Any lymphatic obstruction leads to an enlarged lymph sac and enlarged thoracic duct. This exerts pressure on the heart and displace the heart which results in obstruction of blood flow through the cardiac chambers resulting in abnormal growth of certain cardiac structures.

Prevalence is

- 0.8/1000 when NT is normal
- 28.9/1000 when NT is 3.5 – 4.4
- 90.9/1000 when NT is 4.5 – 5.4
- 195.1/1000 when NT is > 5.5

## **2. Chromosomal abnormalities**

Many of the component proteins of the extracellular matrix are encoded on chromosomes 21, 18 or 13. Immunohistochemical studies, examining the skin of chromosomally abnormal fetuses, have demonstrated specific alterations of the extracellular matrix, which may be attributed to gene dosage effects.

Altered composition of the extracellular matrix may also be the underlying mechanism for ↑ fetal NT in a number of genetic syndromes, abnormalities of fibroblast growth factor receptors or disturbed metabolism of peroxisome biogeneric factor.<sup>40</sup>

### **3. Failure of lymphatic drainage**

Dilatation of jugular lymphatic sac results in increased NT. Dilatation of lymphatic sacs results from delayed development in connection of lymphatics with the venous system which interrupts normal flow between lymphatic and venous system. It may also result from abnormal dilation of lymphatic channels or proliferation of the lymphatic channels.<sup>41</sup>

### **4. Venous congestion in head and neck**

Constriction of fetal body or chest resulting from various condition leads to venous congestion of head and neck.<sup>43</sup>

## **5. Fetal anemia and hypoproteinemia**

Fetal anaemia resulting from genetic causes, infection, can present with heart failure which can present with increased fetal NT.<sup>43</sup>

## **6. Neuromuscular abnormalities**

Causes poor breathing and body movements, which may lead to fluid accumulation as that happens in peripheral edema e.g., arthrogryposis – Contractures and flexion-deformities.<sup>41</sup>

## **7. Fetal infection**

Increased NT is associated with Parvovirus B<sub>19</sub>. Torch group of organisms have less association with increased nuchal translucency. In fetal infections, there is suppression of hemopoiesis due to myocardial dysfunction, fetal anemia which leads to increased NT.

### **Transient nature of Nuchal Translucency between 10-14 weeks**

The fetal lymphatic system is developing between 10-14 weeks of gestation and the peripheral resistance of the placenta is also high. So the excess fluid accumulates behind the neck leading to increased NT. After 14 weeks of gestation, the lymphatic system is well developed to drain away the excess fluid to placental circulation. After this period, fetal abnormalities causing fluid accumulation will be corrected by itself and goes undetected by measuring NT.<sup>30</sup>

Reason for selecting up to 14 weeks for NT:

1. To provide women safe and earlier termination when fetus is affected
2. The incidence of increased NT is lower at 14-18 weeks than at < 14 weeks of gestation in affected fetus.<sup>45, 46, 47</sup>
3. Fetus will be in horizontal position at 10-14 weeks of gestation, ideal position for taking measurement with a success rate of 98% - 100%. It is difficult to obtain appropriate image beyond 14 weeks as the fetus will often be in vertical position.

Reason for selecting 10 weeks as lower limit:

1. CVS was the diagnostic test before. CVS done before 10 weeks was associated with transverse limb reduction defects.
2. Fetal structures are visualized after 10 weeks, any fetal abnormalities can be diagnosed with NT scan.

## **METHODS OF MEASUREMENT OF NUCHAL TRANSLUCENCY(NT):**

NT can be measured by transabdominal or transvaginal ultrasound. Scan should be done by trained Radiologist. The equipment must be of good quality, with 5MHz transabdominal probe or 8MHz transvaginal probe. It should have a video loop junction to 1 decimal point. NT scan should be done for at least 10-20 minutes. NT measurement should be tried for at least 20 minutes before abandoning the procedure. CRL should be correctly measured with fetus in proper sagittal view of spine.<sup>53</sup>

1. Fetus is examined in mid sagittal section, either facing towards or away from the transducer with thalamus, medulla oblongata and pons are visible.
2. Fetus must be in neutral position especially the neck for NT to be measured. Standard Techniques for NT measurement is the Fetal medicine foundation (FMF) criteria.

When the fetal neck is hyperextended NT value increases by 0.6mm and when it is flexed the NT value decreases by 0.4mm.<sup>54,55</sup>

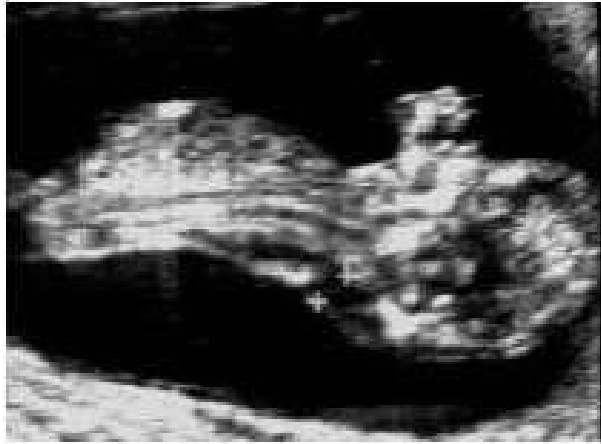




**Fig. 7: Nuchal translucency measurement that meets Fetal Medicinal Foundation criteria (t=thalamus, mo=medulla oblongata, p=pons). The correct caliper placement is “on-to-on”.**



**Fig.8 Magnification is too small for accurate measurement of NT**



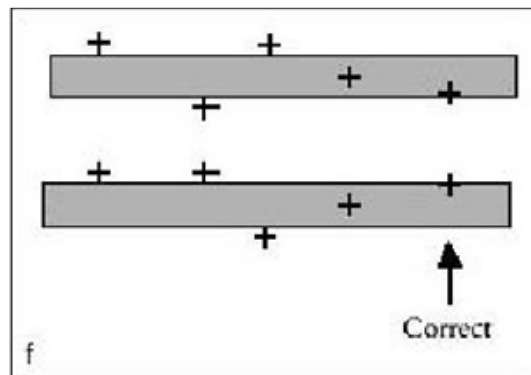
**Fig.9 Neck is hyperextended, not correct for measuring NT**



**Fig.10 Neck is too much flexed, not suitable for measuring NT**

3. The gestational age should be 10-14 weeks and fetal CRL should correspond to 38-84mm. According to fetal medicine update – gestational age should be 11-13+6 weeks and fetal CRL correspond to 45-84mm.<sup>56, 57</sup>

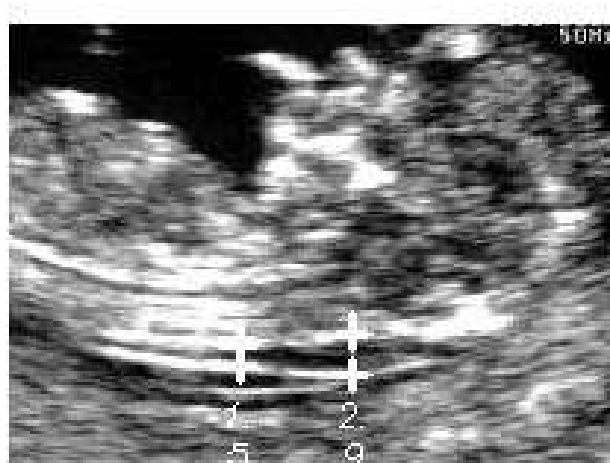
4. Fetal image should occupy 75% of the screen. Fetal image should include only the fetal head and upper thorax.<sup>58</sup>
5. Magnification of the image should be as large as possible, so that slight movement of calipers produces only 0.1mm change in measurement.<sup>58</sup> Caliper should be placed correctly i.e., on inner border of nuchal fold and perpendicular to long axis of the fetal body i.e., “on to on” caliper placement only cross-shaped (+) caliper is used.



**Fig. 11: Correct placement of calipers**

6. The fetal skin and amnion should be distinguished carefully since both structures appear as thin membranes. They are distinguished when there is spontaneous fetal activity, the fetus moves away from amniotic membranes. This is also achieved when the mother coughs or mother's abdomen is tapped.<sup>2</sup>

7. Measurement of maximum thickness of subcutaneous translucency between the skin and the soft tissue overlying the cervical spine is taken. Multiple measurement are taken and maximum one is recorded to be more accurate.<sup>2</sup>



**Fig.12 Maximum measurement of NT to be taken.**

8. In 5% of cases umbilical cord surrounds the fetal neck and this produces falsely increased NT. In this case, NT measurements above and below the cord levels are different, it is best to use the average of those two measurements.



**Fig. 13: Umbilical cord surrounds the fetal neck that disrupts NT measurement.**

**Pitfalls in measurement:**

- Loose amnion that can be mistaken for nuchal skin edge
- Nuchal cord
- Encephalocele
- An amniotic band

**Methods to avoid Bias:**

- To wait for fetal activity to differentiate the amnion from fetal skin
- In nuchal cord - colour Doppler may be helpful
- Adequate magnification distinguishes fetal skin and amniotic membrane

**Per-requisites:**

- 1) Appropriate training
- 2) High motivation
- 3) Strict adherence to FMF technique for measurement

**Implementation of NT screening in routine:****NT – Calculation of patient-specific risk**

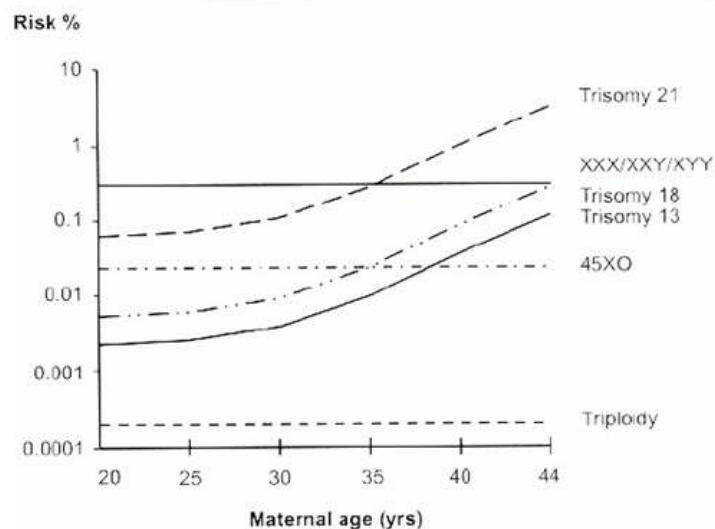
- Every fetus has its own risk to have chromosomal defect. To calculate the individual risk, it is necessary to take into account the background or a “prior risk” that depends on the maternal age and gestation and it is multiplied by a series of factors, resulting from the series of screening tests carried out during the course of the pregnancy.
- Dividing the percentage of chromosomally abnormal fetuses by the percentage of normal fetuses gives the likelihood ratio for a given sonographic/biochemical measurement.

- Prospective studies were conducted among 2,00,000 pregnancies, in which more than 900 fetuses were with Down's Syndrome. This study proved that NT measurement can identify more than 75% of fetuses with Down's Syndrome with a false positive rate of 5%.<sup>64</sup>

## OTHER SCREENING MODALITIES FOR CHROMOSOMAL DEFECTS

### 1. Maternal Age:

In 1909, Shuttleworth was the first to notice that mothers of Down's children were of older age than average. Chromosomally abnormal fetuses are more likely to die in utero than normal fetuses, so the risk decreases with gestational age and increases with increasing maternal age.



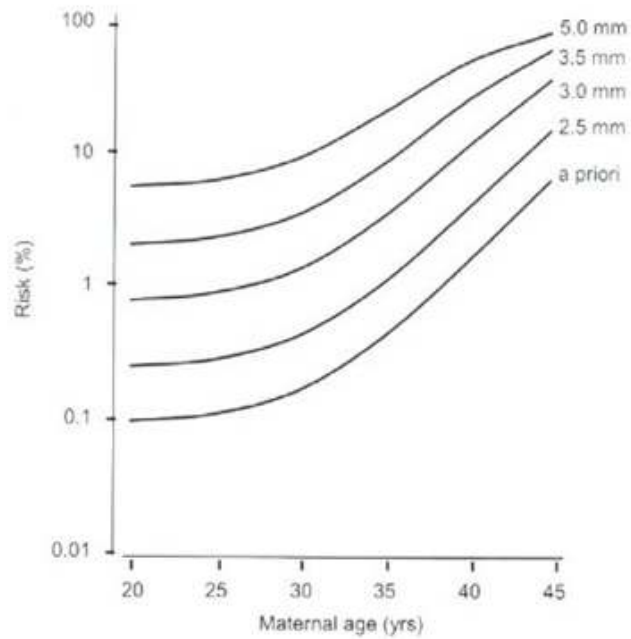
**Fig. 14: Maternal age related for chromosomal abnormalities**

Previously screening for Down's Syndrome was based on the advanced maternal age. When screening conducted on this basis only 30% of down's babies was detected.<sup>34</sup>



**Estimated risk for trisomies 21, 18, and 13 in relation to  
maternal age and gestation<sup>68</sup>**

<b>Maternal Age (Years)</b>	<b>Trisomy 21</b>				<b>Trisomy 18</b>				<b>Trisomy 13</b>			
	<b>Gestation (wks)</b>				<b>Gestation (wks)</b>				<b>Gestation (wks)</b>			
	12	16	20	40	12	16	20	40	12	16	20	40
20	1068	1200	1295	1527	2484	3590	4897	18013	7826	11042	14656	42423
25	946	1062	1147	1352	2200	3197	4336	15951	6930	9778	12978	37567
30	626	703	759	895	1456	2013	2869	10554	4585	6470	8587	24856
31	543	610	658	776	1263	1825	2490	9160	3980	5615	7453	21573
32	461	518	559	659	1072	1549	2114	7775	3378	4766	6326	18311
33	383	430	464	547	891	1287	1755	6458	2806	3959	5254	15209
34	312	350	378	446	725	1047	1429	5256	2284	3222	4277	12380
35	249	280	302	356	580	837	1142	4202	1826	2576	3419	9816
36	196	220	238	280	456	659	899	3307	1437	2027	2691	7788
37	152	171	185	218	354	512	698	2569	1116	1575	209	6050
38	117	131	142	167	272	393	537	1974	858	1210	1606	4650
39	89	100	108	128	208	300	409	1505	654	922	1224	3544
40	68	76	82	97	157	227	310	1139	495	698	927	2683
41	51	57	62	73	118	171	233	858	373	526	698	2020
42	38	43	46	55	89	128	175	644	280	395	524	1516



**Fig. 15: Maternal age related for trisomy 21 at 12 weeks of gestation and the effects of fetal nuchal translucency thickness**

In 1990, screening for Trisomy 21, with maternal age was combined with fetal NT thickness at 10-14 weeks of gestation. This improved the detection rate to 75% of affected fetuses with Trisomy 21.

## **2. Other ultrasound markers in I trimester:**

In the I trimester, presence of nasal bone, measurement of fronto maxillary facial angle, Doppler study of tricuspid valve and ductus venosus, any major structural anomalies are performed.

### **Nasal Bone:**

Nasal bone is visualized by ultrasound done in first trimester and the position of fetus to visualize the nasal bone is same as NT measurement. The face of transducer is parallel to long axis of the nasal bone and the skin over nasal bridge.

The skin over the nasal bridge and the nasal tip visualized as echogenic lines. The nasal bone present within the substances of the nasal bridge is also seen as echogenic line. When nasal bone is present, “equal sign” is present.



**Fig. 16: (16a) The nasal bone (NB) is present (solid arrow). The NB and the echogenic skin over the nasal bridge (open arrow) create then so called “equal sign”.**



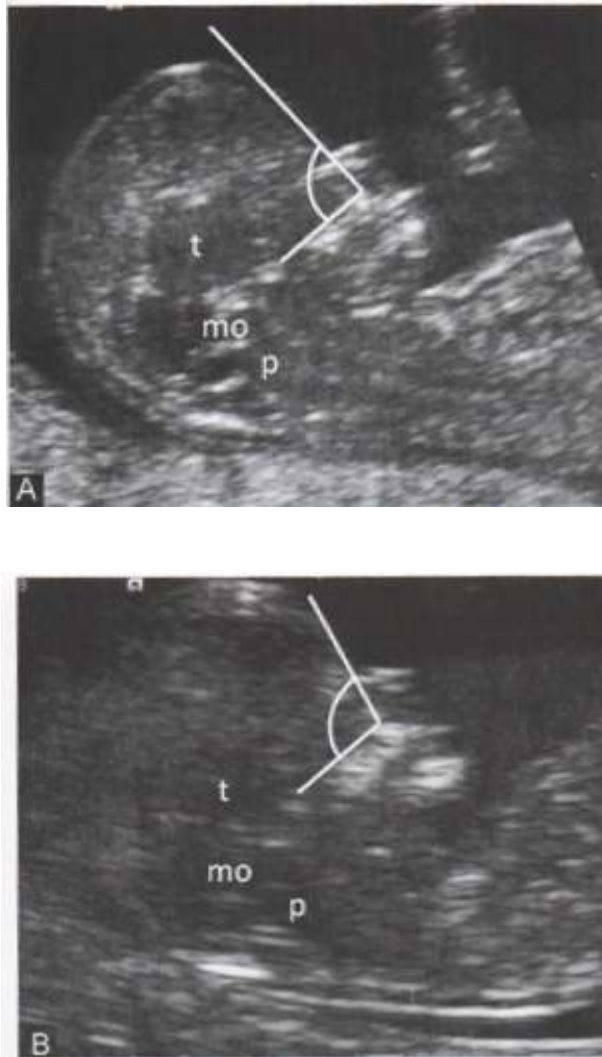
**Fig.(16b) Nasal bone is absent (note absence of the “equal sign”. Only a single echogenic line representing the echogenic skin over the nasal bridge (open arrow) is present. (T=thalamus, BS=brain stem)**

Appearance of nasal bone is based on the facial features in individuals with Down's syndrome. There may be difference in size or degree of ossification of nasal bone or the nasal bone may be absent. This may be due to changes in connective tissue that happens in Trisomy 21.

Nasal bone is absent in approximately 60% of fetuses with Trisomy 21 and 1-2.5% of euploid fetuses. NB evaluation when combined with first trimester screening led to increased detection rate to 90%.

**Frontomaxillary facial angle (FMF angle):**

The fetus is in the same position of NT measurement and FMF angle is measured using Fetal Medicine Foundation criteria. First ray is drawn along the upper edge of hard palate, apex is on the anterior aspect of maxilla, second ray is from apex upwards resting on echogenic line beneath the skin (non-calcified metopic suture). FMF angle is acute in normal conditions.



**Fig. 17A and B: Frontomaxillary angle measurement that meets Fetal Medicine Foundation criteria (t=thalamus, mo=medulla oblongata, p=pons). Note the absence of the frontal process of the maxilla between the hard palate and the nasal bone, which helps to assure that plane of insonation is precisely in the midline. (A) The frontomaxillary angle is acute and within normal limits: (B) The frontomaxillary angle is obtuse and is above the normal range**

Flat faces is a common dysmorphic feature in Down's babies due midface hypoplasia resulting from abnormal connective tissue. This flatness of the face is assured by FMF angle.

When performed on 3D volumes demonstrated that 70% of Down's syndrome fetuses had FMF angle measurement that was above 95<sup>th</sup> percentile of the normal population. In 2D technology, this rate falls to 50%. FMF angle is independent of NT measurement, nasal bone and serum biochemistry.

### **Intracranial Translucency (IT) :**

Intracranial Translucency is another sonographic marker of fetal anomaly especially spina bifida. IT is measured at 11-13 weeks of gestation, with the fetus in mid sagittal view and in neutral position and magnification same as in NT measurement .

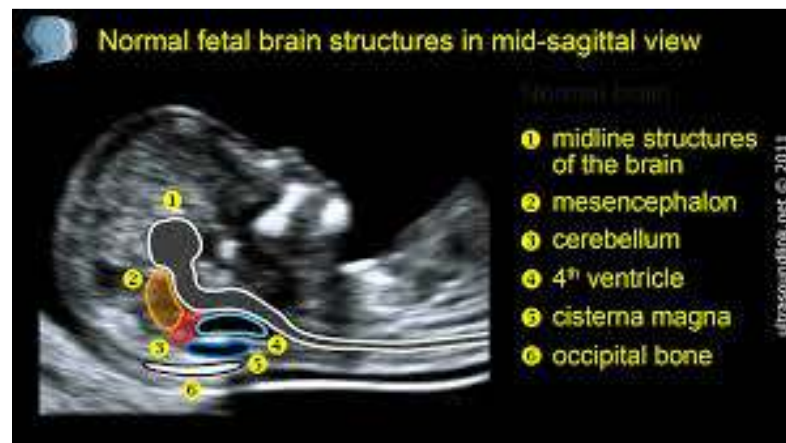


Fig 18(A). Fetal brain structures in mid sagittal view and 4<sup>th</sup> ventricle represents the Intracranial Translucency.

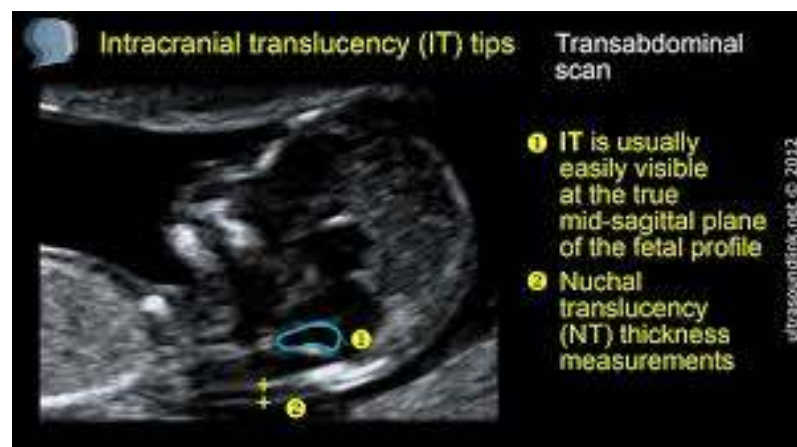


Fig 18(B). Intracranial Translucency measurement.



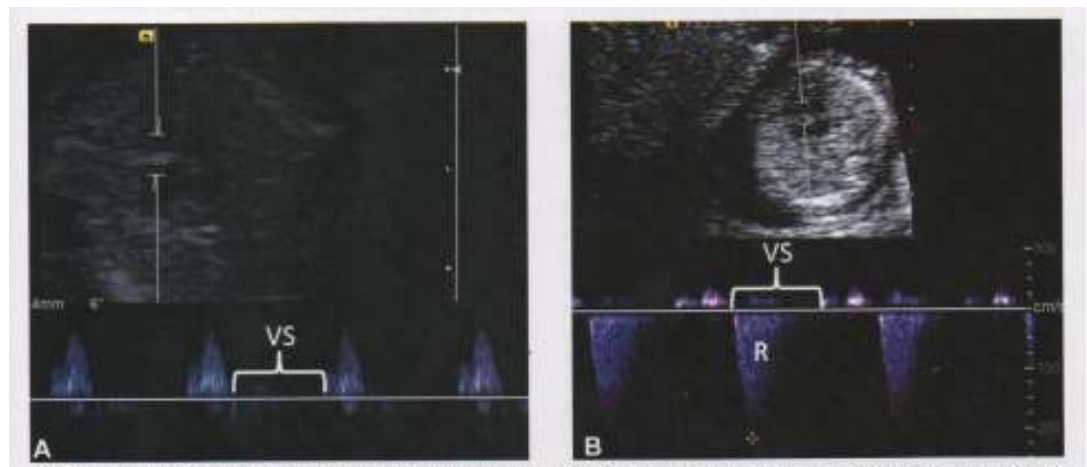
Intracranial Translucency is the measurement of the fourth cerebral ventricle between the brain stem and the choroid plexus. IT is measured as anteroposterior diameter. In spina bifida, there is caudal displacement of hindbrain which compresses the fourth ventricle. This space which was measured as IT will be obliterated due to this displacement. If IT is not measurable, the fetus is at risk of spina bifida especially open spina bifida.

**Doppler evaluation of fetal blood flow as markers for aneuploidy:**

There is high interventricular pressure at any cardiac volume due to lower compliance of fetal myocardium. Placental vascular resistance is high in early pregnancy causing additional strain on the heart. Any abnormalities of cardiac structures or performance leads to detectable changes in blood flow through Tricuspid valve and Ductus venosus.

### **Tricuspid valve regurgitation:**

In Trisomy 21, there is increased prevalence of tricuspid valve regurgitation may be due to decreased number of myocytes with decreased orientation of the myocytes and abnormal connective tissue. It may also result from dilatation of right ventricle with dilatation of tricuspid valve annulus or connective tissue abnormality of the valve itself.



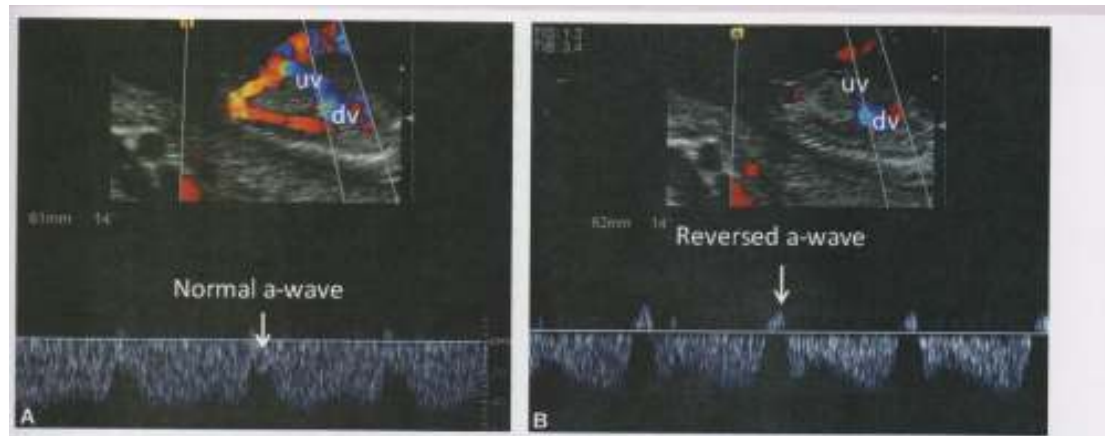
**Fig 19A and B. Evaluation of the flow across the tricuspid valve (TCV) using Doppler; (A) Normal flow pattern across the TCV with no regurgitant flow during ventricular systole (VS); (B) Abnormal flow pattern across the TCV with regurgitant flow (R) present during ventricular systole (VS)**

Normal tricuspid valve waveform shows Biphasic pattern of blood flow into the right ventricle. First wave is the increase in forward flow at the beginning of ventricular diastole followed by a decrease. Second wave is increase in flow corresponding to atrial systole. Any flow across the tricuspid valve during ventricular systole represents regurgitation. In tricuspid valve regurgitation flow across the valve must be atleast 30% of ventricular systole and the peak velocity of regurgitation jet must be more than 60cm/sec.

At 11-14 weeks of gestation, prevalence of tricuspid valve regurgitation in Trisomy 21 fetuses is 55%, in euploid fetuses it is 1%, in Trisomy 18 fetuses it is 33%, in Trisomy 13 it is 30% and monosomy 38%.

#### **Ductus venosus – Reversed A wave:**

Normal waveform demonstrates forward flow throughout the cardiac cycle with two adjoining peak of increased flow in ventricular systole and diastole, and a decreased flow during atrial systole but forward flow is maintained. 'A' wave represents atrial systole.



**Fig 20A and B. Evaluation of ductus venosus (DV) flow using Doppler (uv=hepatic portion of the umbilical vein) (A) Normal DV flow pattern with antegrade flow during the entire cardiac cycle, including during the atrial contraction (a-wave); (B) abnormal DV flow pattern with reversed flow during atrial contraction(a-wave).**

Reversed 'A' wave in ductus venosus Doppler occurs in aneuploidies which results from decreased compliance of ventricular wall rather than ventricular dilation. Abnormalities in 'A' wave are also associated with increased risk of cardiac anomalies and it is followed up by second trimester target scan and fetal echocardiography.

The prevalence of reversed flow of 'A' wave in Trisomy 21 is 66%, Trisomy 18 is 58%, Trisomy 13 is 55%, monosomy is 75% and in euploid is 3%.

### **Fetal Heart Rate (FHR) screening for aneuploidy:**

Aneuploidy fetuses have different heart rates at first trimester compared with normal fetuses. In Trisomy 21, 14% of fetuses have fetal heart rate >95<sup>th</sup> percentile. In Trisomy 13 and monosomy, fetus have FHR > 95<sup>th</sup> percentile in 69% and 53% respectively. In Trisomy 18 and triploidy, fetuses tend to be bradycardiac. FHR needs to be adjusted for gestational age when included in first trimester screening.

### **Screening for fetal structural anomalies in I trimester:**

Structural anomalies like holoprocencephaly, diaphragmatic hernia, atrioventricular septal defect, omphalocele, megacystis are associated with aneuploidies and are detectable in I trimester by ultrasonogram.

### **3. Maternal serum screening in I trimester:**

Now a days it is noted that PAPP-A (Pregnancy associated Plasma Protein – A) is probably the best marker for Down's syndrome in I trimester.

PAPP-A is a pregnancy specific glycoprotein produced by the trophoblast. It becomes detectable in maternal serum from 28 days after conception. It is low in Down's syndrome and other trisomies.

Free  $\beta$ -HCG is another biochemical marker used in Down's syndrome. It is elevated in Trisomy 21 and other trisomies.

- Maternal age + PAPP-A » detects 71% cases of Down's syndrome
- Maternal age + PAPP-A + free  $\beta$ -HCG » detects 85-90% of Down's syndrome

**I trimester screening for aneuploidy using multiple ultrasound markers – Risk assessment by Fetal Medicine Foundation:**

This risk assessment is done to categorize the women at risk for aneuploidy into high risk, intermediate risk and low risk in the first trimester itself.

This approach includes the following parameters

- Maternal Age
- NT measurement
- Free  $\beta$ -HCG
- PAPP-A level

Includes 3 risk categories depending on the risk of Trisomy 21

**1. High Risk:**

Women whose risk of trisomy 21 is 1:50 or greater.

They are offered invasive testing like amniocentesis / chorion villous sampling

**2. Intermediate risk:**

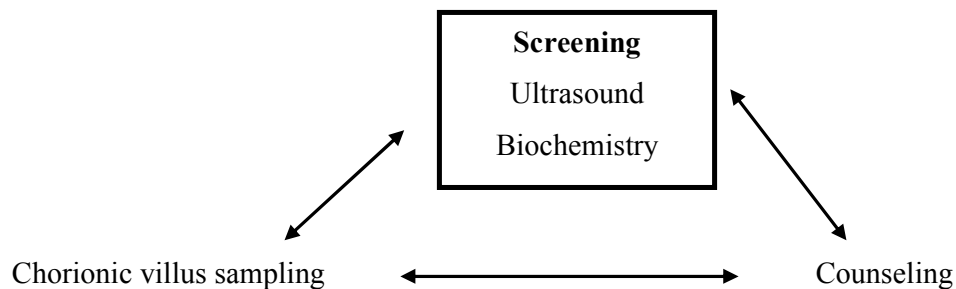
- Women with risk 1:51 to 1:1000
- They are assessed with additional markers like Nasal bone, Doppler study of tricuspid valve and Ductus venosus.
- After additional assessment, if they fall in high risk » invasive testing is done.
- If they fall in low risk » followed up with targeted ultrasound at 20 weeks

### 3. Low Risk:

- Women with risk less than 1:1000
- No testing is done
- Followed up with targeted scan at 20 weeks

### OSCAR (One Stop Clinic For Assessment of Risk)<sup>77</sup>

Recently, maternal age was combined with fetal NT and maternal Serum biochemistry (free  $\beta$  HCG +PAPP-A) in I trimester to identify about 85-90% of affected fetus. Furthermore, the development of new methods of biochemical testing, within 30 minutes of taking a blood sample, made it possible to introduce One - Stop Clinic for Assessment of Risk.



The nasal bone will not be visible in I trimester scan in 60-70% of fetuses with Trisomy 21. So I trimester scan along with serum Biochemistry will detect more than 95% of Trisomy 21.<sup>78</sup>



#### **4 Invasive testing in first trimester:**

Invasive testing available in first trimester are  
Chorionic villus sampling (CVS) and Celocentesis.

##### **Chorionic villus sampling (CVS):**

CVS is performed either transcervically or transabdominally. It is an invasive procedure used for fetal karyotyping and DNA analysis prenatally. Previously CVS was done at 7-8 weeks which had higher incidence of limb defects. Now CVS is done at or after 10 weeks to reduce the fetal defects caused by the procedure.

Indication:

CVS is done for mainly for the following indications.

##### **1.Monogenic disorders- Thalassemia**

Cystic fibrosis

Hemophilia

Duchene muscular dystrophy

Becker's muscular dystrophy

##### **2.Metabolic disorders- Mucopolysacharidosis**

Lipidosis

Aminoacid metabolic disorders

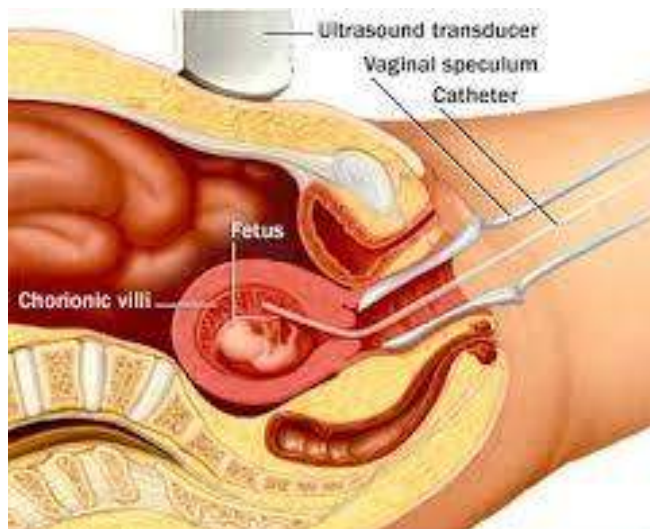
Carbohydrate metabolic disorders

Technique:

CVS is performed either transcervically or transabdominally.

Transcervical CVS:

It is performed with polyethylene catheter or biopsy forceps under the guidance of real time ultrasound. Pregnant women in lithotomy position, under strict aseptic precaution , the aspiration catheter or biopsy forceps is advanced transcervically to obtain the sample. A sample of 5-40mg is sufficient for prenatal diagnosis.

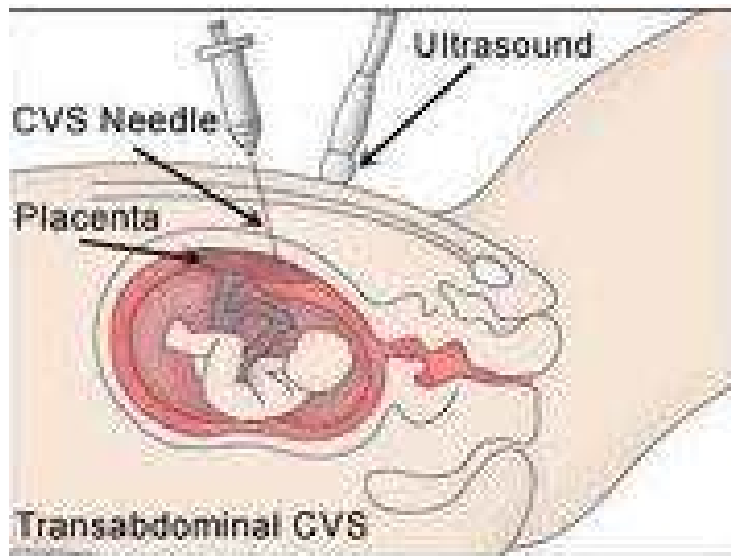


**Fig 21. Transcervical chorionic villus sampling**

### Transabdominal CVS:

It is performed with aspiration needle either by the “Two needle” or the “free hand” (single needle) technique. Pregnant women in supine position under strict aseptic precaution, the aspiration needle is used to aspirate 5-40mg of tissue. This is technically similar to mid trimester amniocentesis.

Transabdominal CVS will be difficult in posterior placental site, potential bowel adhesions, retroversion and retroflexion of uterus which needs transcervical approach.



**Fig 22. Transabdominal chorionic villus sampling.**

CVS allows early diagnosis. Genetic results will be available within hours by direct preparation of the cytotrophoblast layer or within 3-7 days by tissue culture of chorionic villus mesenchymal core.

Complications:

- CVS done before 10 weeks led to fetal limb reduction defects. This is reduced by performing CVS at or after 10 weeks of gestation.
- Fetal loss is another complication, but it is similar to that of amniocentesis.
- Vaginal bleeding
- Post CVS infections

**Celocentesis:**

This is another method of invasive procedure for screening fetal anomalies screening. It is done at 7-8 weeks of gestation which is done earlier than CVS.

Indications:

- To determine fetal sex
- $\beta$  thalassemia
- Sickle cell anemia

- Marfans syndrome
- Paternity testing

Technique:

Celomic cavity occupies most of the gestational sac till 9 weeks of gestation. Celocentesis is performed transvaginally by inserting a 20G needle into the celomic cavity under ultrasound guidance. 1-2 ml of the celomic fluid is aspirated and sent for biochemical analysis and cytogenetic studies.

Complications:

- Fetal loss
- Fetal limb reduction syndrome
- Fetal oromandibular hypoplasia
- Intracelomic hemorrhage

### **Maternal Serum Screening in II trimester:**

A new method of screening was introduced in 1980 that considers maternal age and the concentration of various fetoplacental biochemical products in the maternal circulation.<sup>69</sup>

The maternal serum concentration of AFP ( $\alpha$  Fetoprotein), unconjugated estriol (UE3), human chorionic gonadotrophin(HCG) (total and free  $\beta$ ) are measured at 16 weeks of gestation. In Trisomy 21  $\beta$ -HCG will be elevated UE3 and AFP will be decreased. These 3 serum markers are used in triple test. Another marker Inhibin A is used along with triple test which is elevated in aneuploidy. This concept is used to identify the high risk group.

	Detection Rate of Trisomy21
MSAFP	36%
Dual Test – MSAFP + Free $\beta$ HCG	58%
Triple Test- unconjugated Estriol + $\beta$ -HCG + AFP	67%
<div>↓                  ↑                  ↓</div>	
Quad Test - Triple test + Inhibin A ( $\uparrow$ ) <sup>71,72</sup>	70%

## **Invasive testing in II trimester:**

Amniocentesis and Cordocentesis are invasive procedures done in II trimester.

### **Amniocentesis :**

Amniocentesis is the invasive testing available for screening in the II trimester and it is a safe technique among the invasive procedures. Amniocentesis was first done therapeutically for polyhydramnios to drain excess amniotic fluid. Later it was used for Rh isoimmunisation, then for sex identification and now it is used as a screening test for fetal chromosomal abnormalities and other congenital defects.

Indications:

- Fetal karyotyping
- Genetic diseases- $\beta$  thalassemia

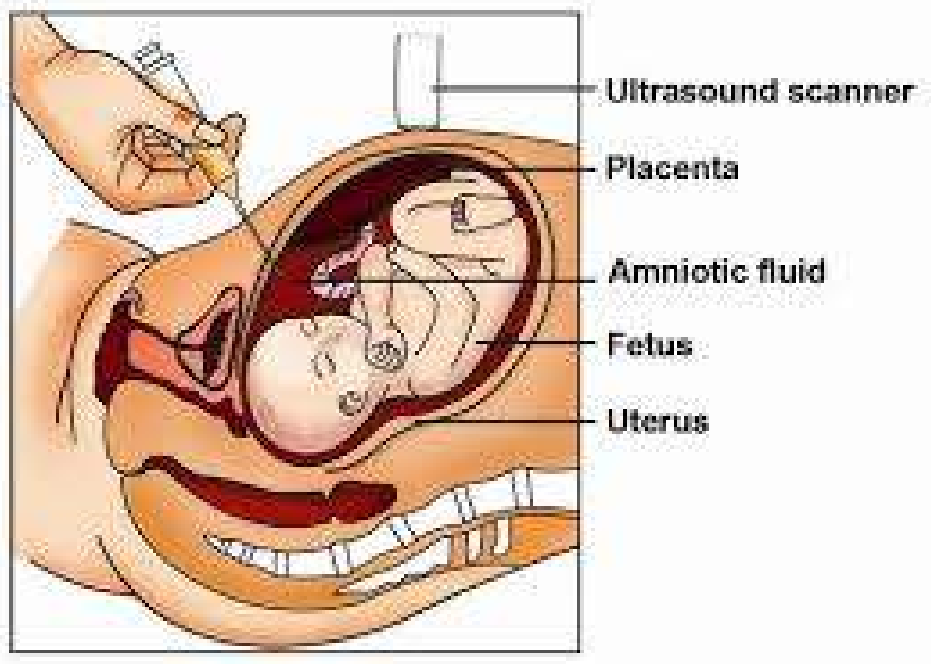
Cystic fibrosis

Hemophilia

Congenital adrenal hyperplasia

Fetal lung maturity

- Congenital fetal infections-TORCH
- Severity of fetal hemolytic anaemia in Rh isoimmunisation
- Fetal therapy in twin oligohydramnios - polyhydramnios sequence and twin-to-twin transfusion syndrome
- Amnioinfusion in oligohydramnios
- Infusion of drugs into amniotic cavity



**Fig 23. Amniocentesis under ultrasound guidance.**



### Technique:

Amniocentesis is performed at 15-18 weeks of gestation. It is performed under strict aseptic precaution with 22G needle under direct ultrasound guidance. Detailed ultrasound examination of the fetal anatomy, amniotic fluid volume, placental location, uterine abnormality and adnexal masses are done prior to the procedure. The site of entry of the needle must be away from the face, umbilical cord and placenta. 20 ml of the fluid is aspirated and sent for cytogenetic study. Repeat ultrasound is done to confirm fetal cardiac activity and absence of intraamniotic bleeding.

In twin pregnancies double amniocentesis is done with either one or two separate 22G needles. When two needles are used, they are inserted sequentially in each sac and 20ml of amniotic fluid is aspirated from each sac. When one needle is used, the needle is entered into the proximal sac near the insertion of the dividing membrane and 20ml of amniotic fluid is aspirated. Then the needle is advanced through the second sac under direct ultrasound guidance and 20 ml of amniotic fluid is aspirated from the second sac.

### Complications:

- Fetal loss
- Fetal injury
- Preterm delivery
- Rh isoimmunisation from fetomaternal hemorrhage
- Neonatal respiratory distress syndrome due to postprocedure oligohydramnios
- Orthopedic abnormalities due to oligohydramnios
- Vaginal spotting
- Leakage of amniotic fluid.

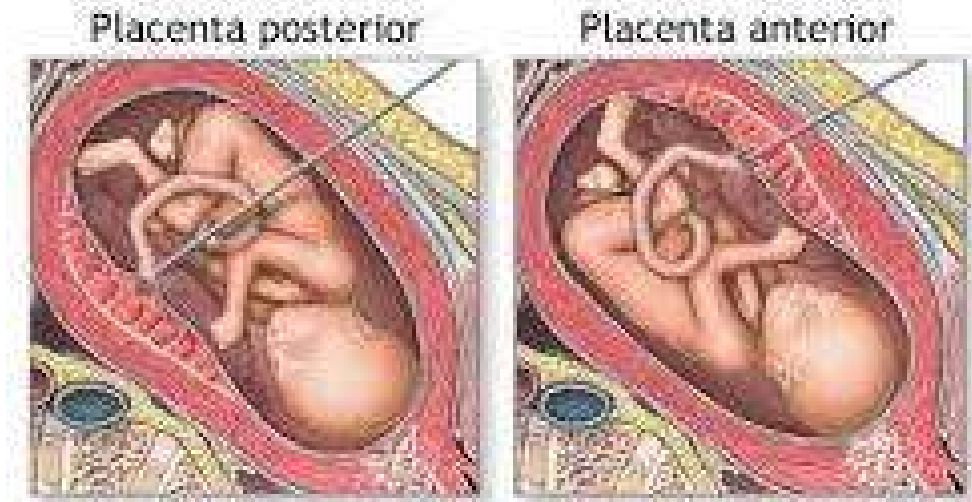
### **Cordocentesis / Percutaneous Umbilical Blood Sampling(PUBS):**

Cordocentesis is done transabdominally under ultrasound guidance. This procedure has been limited by CVS and amniocentesis. PUBS has been done mostly for confirmation of abnormal findings in amniocentesis and CVS.

### Indications:

- Rh isoimmunisation
- Hydrops of unknown origin
- Fetal infections

- To inject drugs into fetal circulation
- For blood transfusion in fetal anaemia



**Fig 24. Transabdominal ultrasound guided fetal blood sampling.**

Technique:

A detailed ultrasound examination of the fetus, placenta, uterus are done. Under strict aseptic precaution, with ultrasound guidance, a 22G needle is introduced either trans-placentally in case of anterior placenta or through the amniotic cavity in case of posterior placenta targeting the umbilical vein proximal to the insertion of umbilical cord into the placenta. The intrahepatic portion of the umbilical vein, the left portal vein and the right ventricle have been used to obtain fetal blood sample.

4ml and 6ml of fetal blood should be withdrawn for sampling during the second and third trimester respectively. The cord puncture site should be followed up sonographically for 10 minutes for bleeding or hematoma and fetal heart rate for 30-60 minutes after the procedure.

Complications:

- Fetal loss
- Bleeding at puncture site
- Umbilical cord hematoma
- Transient fetal bradycardia
- Fetomaternal blood transfusion
- Uterine contractions
- Chorioamnionitis

<b>Method of Screening<sup>34</sup></b>	<b>DR% of Down Syndrome</b>
Material Age (MA)	30%
MA + Material serum Biochemistry at 15-18 weeks	50-70%
MA+ Fetal NT	70-80%
MA+ Fetal NT +serum free $\beta$ HCG+PAPP-A at 10-14 weeks	85-90%
MA +Fetal nasal bone + Fetal NT	90%
MA + Fetal NT+ NB+ Maternal serum	95%
Free $\beta$ HCG +PAPPA at 10-14 weeks	

## **II Trimester Ultrasonography<sup>17</sup>**

Benacerraf's scoring system for Down Syndrome:

<b>Findings</b>	<b>Score</b>
Major Anomaly	2
Nuchal fold $\geq 6$ mm	2
Hyperechoic bowel	1
Echogenic intracardiac focus	1
Pyelectasis $\geq 4$ mm	1
Short femur	1
Short humerus	1

**Note:** A score of  $\geq 2$  warrants an invasive testing.

## **Central Nervous System**

- Holoprocencephaly
- Dandy walker Syndrome
- Agenesis of corpus callosum
- Ventriculomegaly
- Choroid plexus cyst
- Dilated cisterna magna

## **Cardiovascular system**

- VSD
- ASD
- Tetralogy of Fallot's
- Endocardial cushion defects

## **Gastrointestinal system**

- Duodenal atresia
- Omphalocele
- Echogenic bowel
- Diaphragmatic hernia

## **Genitourinary system**

- Pylectasis
- Multicystic dysplastic kidney
- Hydronephrosis

## **Face**

- Harelip, cleft palate
- Ocular and ear anomalies
- Abnormal profile

## **Limbs**

- Limb length discrepancy
- Clubfoot
- Rocker bottom foot
- Sandal toe
- Simian Crease
- Syndactyly

## **Miscellaneous**

- Cystic hygroma
- Non-immune hydrops
- IUGR
- Abnormally large placenta/small placenta
- Poly/oligohydramnios



## *MATERIALS AND METHODS*

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## **METHODOLOGY**

### **Design:**

This study was conducted at Coimbatore Medical College Hospital. This prospective study has been conducted on 50 pregnant women, between 10-14wks of gestation with regular menstrual cycle and with reliable LMP details attending the antenatal Out Patient Department at Coimbatore Medical College Hospital during a period of 1 year from August 2013 – August 2014.

### **Inclusion criteria:**

- Study of 50 pregnant women with reliable LMP details.
- Singleton viable intrauterine pregnancy.

### **Exclusion Criteria:**

- Pregnant women with unreliable LMP details
- Multiple pregnancies

**Method:**

The patients are randomly selected. They are offered counseling about the study and are motivated to attend Antenatal Outpatient Department at the specified time for screening and follow-up. The necessities of screening at each trimester were emphasized. The need for I trimester scan to diagnose the viability, to detect multiple pregnancy, for accurate dating of pregnancy, to assess the possibility for Down's Syndrome, other chromosomal abnormalities, other fetal anomalies by measuring nuchal translucency were explained.

The importances of II trimester scan at 20-22 weeks to detect fetal structural abnormalities, growth of the fetus was explained. The cardiac anomaly detected by fetal echocardiography done at 22-24 weeks was explained. After counseling the patient, screening was offered to the pregnant women.

The patient's detailed history was taken and any risk factor contributing to have fetal abnormality was noted. Thorough clinical examination was made. General examination includes Blood pressure, pulse, presence of pallor, icterus, pedal edema. Systemic and obstetric examination was made in detail. Preliminary investigations were carried out at 10-14 weeks scan, fetal CRL, NT, any obvious structural anomaly in the fetus, uterine anomaly, adnexa, cervix and internal os were noted.

After the scan, the estimated risk of Down's syndrome or other fetal abnormalities are discussed with the patient and her family. If she is at high risk, she is counselled and advised to wait for the target scan and fetal echocardiogram.

1. In target scan, fetus is examined thoroughly for anomalies.
2. Any anomalies present, it is discussed with the patient. In trivial pathology and correctable disorder patient is counseled and advised to continue the pregnancy.

3. If major anomalies are present in the patient is advised for termination of pregnancy.

Patient is followed up till delivery, advised for institutional delivery. If NT > 95<sup>th</sup> centile for gestational age, she was considered to be at high risk.

## *RESULTS*

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## RESULTS

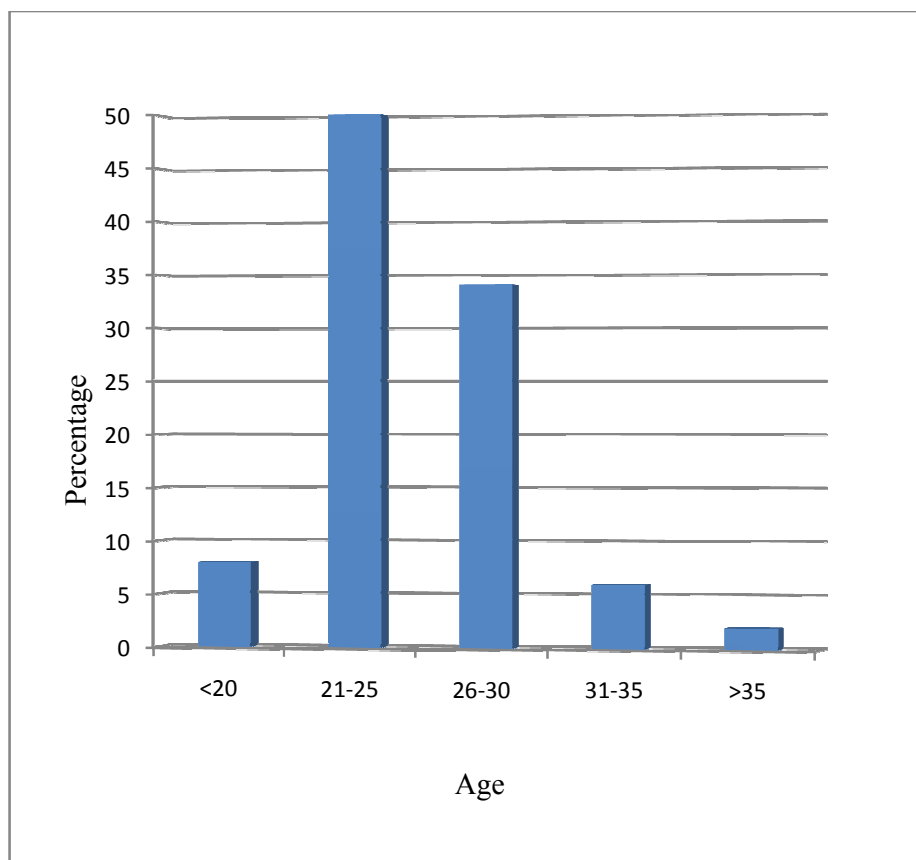
A prospective clinical study consisting of 50 pregnant women is undertaken to study the fetal nuchal translucency at 10-14 weeks of gestation as a screening tool to identify fetal abnormalities.

**Table 1**

### **Age distribution**

<b>Age in years</b>	<b>Number</b>	<b>%</b>
<20	4	8
21-25	25	50
26-30	17	34
31-35	3	6
>35	1	2

**Fig 25: Age distribution**



Most of the pregnant women were between 21-25 years (50%), 34% were between 26-30 years, 8% were between <20 years, 6% were between 31-35 years and 2% were >35 years.

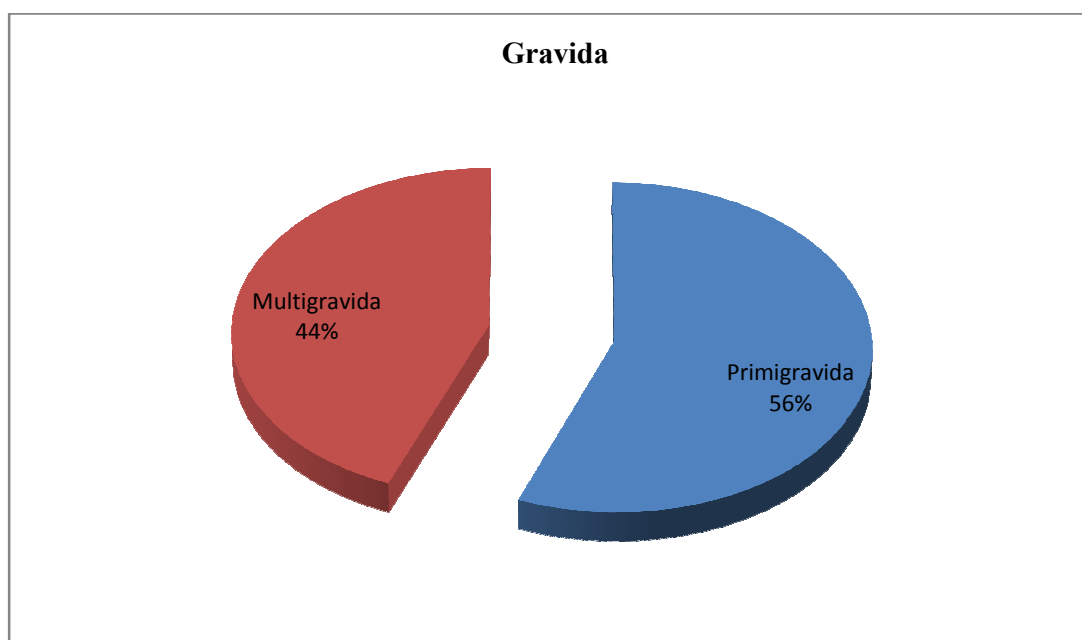


**Table 2**

**Gravida distribution**

Gravida	Number	%
Primigravida	28	56
Multigravida	22	44

**Fig 26: Gravida distribution**



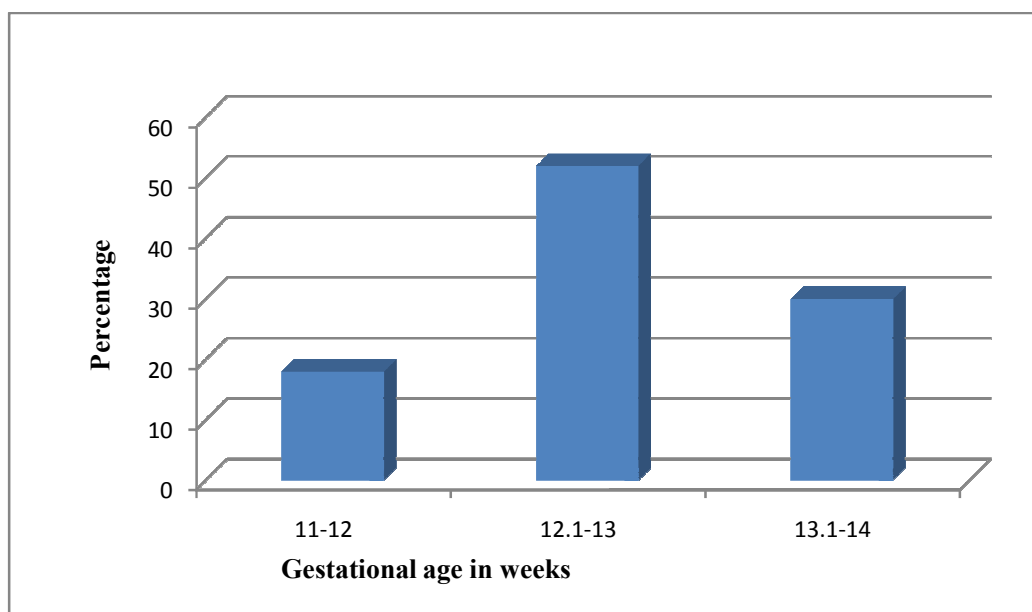
Primigravida were 56% and Multigravida were 44%.

**Table 3**

**Gestational age**

<b>Gestational age in weeks</b>	<b>Number (n=50)</b>	<b>%</b>
11-12	9	18
12.1-13	26	52
13.1-14	15	30

**Fig 27: Gestational age**



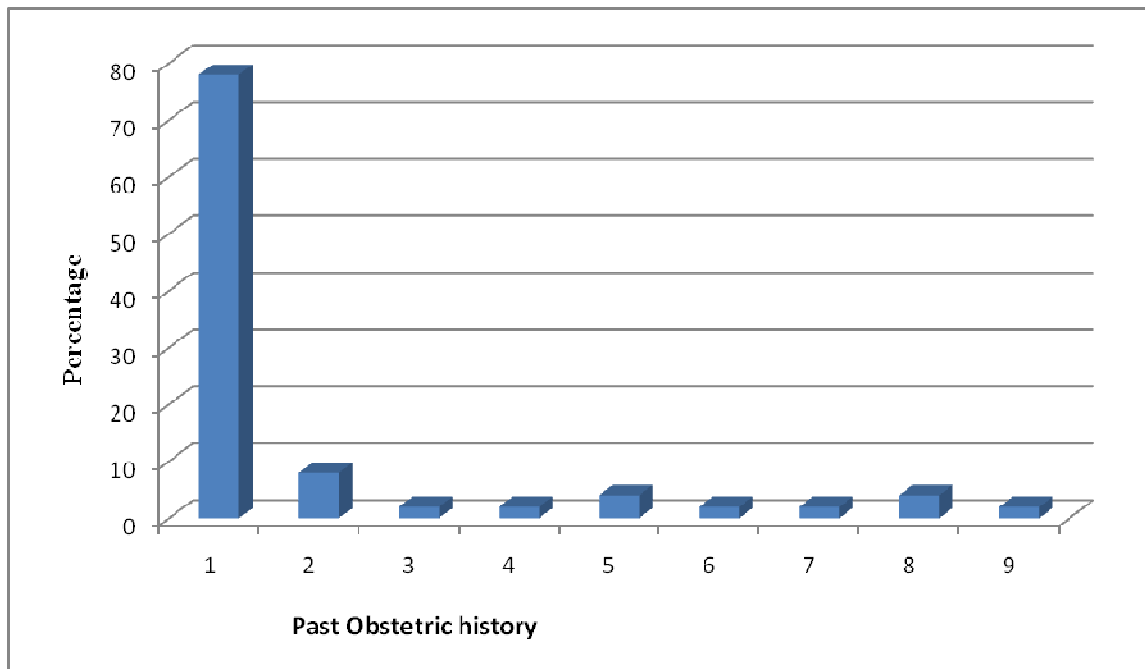
Most of the pregnant women were scanned between 12.1-13 weeks (52%), 30 % between 13.1-14 weeks and 18% were scanned between 11-12 weeks.

**Table 4****Past obstetric history**

<b>Past obstetric history</b>	<b>Number</b>	<b>%</b>
1.Nil	39	78
2.Previous history of unexplained I trimester pregnancy loss	4	8
3.Previous history of unexplained term IUD	1	2
4.Previous history of baby having anomalies	1	2
5.Previous history of unexplained II trimester pregnancy loss	2	4
6.Previous history of preterm delivery	1	2
7.Previous history of IUGR	1	2
8.Previous history of GDM	2	4
9.Previous history of GHT	1	2

One patient had both IUGR and GHT in previous pregnancy. Another patient had I trimester loss and GDM in previous pregnancy.

**Fig 28: Past obstetric history**



78% of pregnant women had no significant risk factors in the past obstetric history.

8% of pregnant women had previous history of unexplained I trimester pregnancy loss.

2% of pregnant women had previous history of unexplained II trimester pregnancy loss.

2% of pregnant women had previous history of unexplained term IUD.

2% of pregnant women had previous history of baby having anomaly.

2% of pregnant women had previous history of preterm delivery.

2% of pregnant women had previous history of IUGR.

4% of pregnant women developed GDM in previous pregnancy.

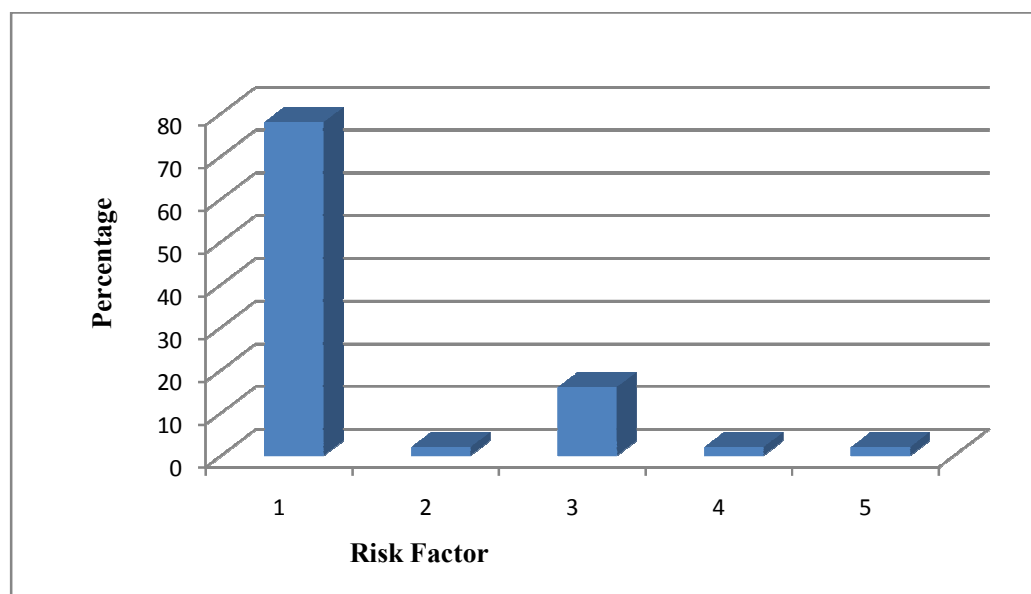
2% of pregnant women developed GHT in previous pregnancy.

**Table 5**

**Risk factor in the present pregnancy identified at the time of NT scan**

<b>Risk factor in the present pregnancy</b>	<b>Number</b>	<b>%</b>
1.Nil	39	78
2.Overt Diabetic	1	2
3.Previous LSCS	8	16
4.Elderly Primi	1	2
5.Hypothyroidism	1	2

**Fig 29: Risk factor in the present pregnancy identified at the time of NT scan**



78% of pregnant women had no risk factor at the time of NT scan. 2% were overt diabetic, 16% were previous LSCS cases, 2% elderly primi, 2% had hypothyroidism.

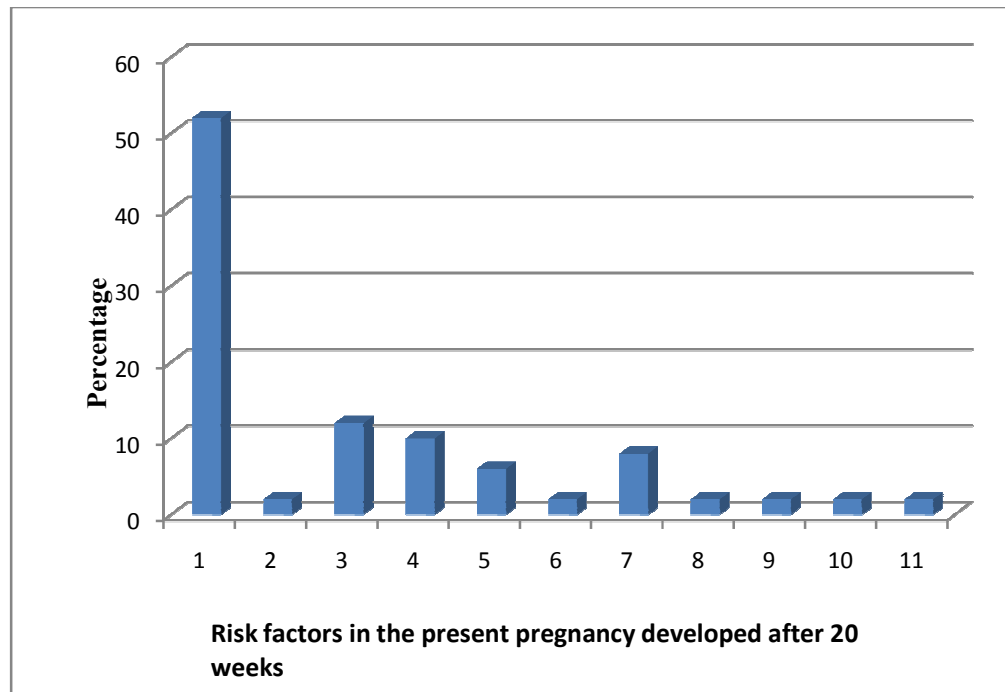
**Table 6**

**Risk factors in the present pregnancy developed later**

<b>Risk factor in the present pregnancy</b>	<b>Number</b>	<b>%</b>
1. Nil	26	52
2. GDM	1	2
3. PIH	6	12
4. Postdated pregnancy	5	10
5. Decreased liquor	3	6
6. Breech presentation	1	2
7. PROM	4	8
8. PPRM	1	2
9. Polyhydramnios	1	2
10. IUGR	1	2
11. Preterm labour	1	2



**Fig 30: Risk factors in the present pregnancy developed later**



52% of pregnant women did not develop any risk factor.

2% of pregnant women developed GDM.

12% of pregnant women developed GHT.

10% of pregnant women went in for postdatism.

6% of pregnant women developed Oligohydramnios.

2% of pregnant women had breech presentation.

8% of pregnant women developed PROM.

2% of pregnant women developed PPRM.

2% of pregnant women developed polyhydramnios.

2% of pregnant women developed IUGR.

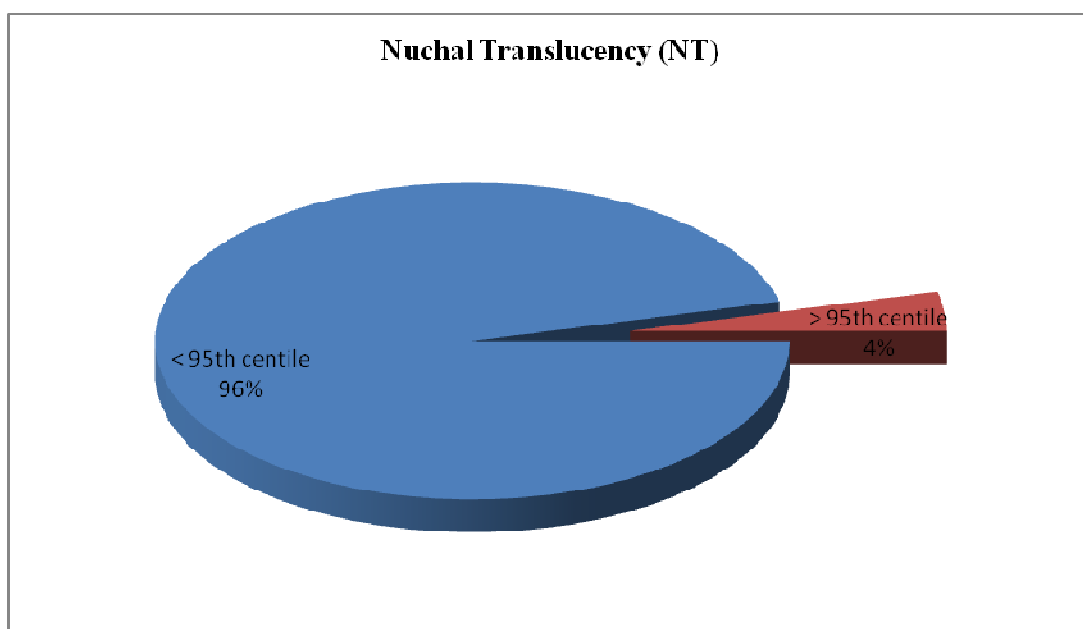
2% of pregnant women had preterm labour

**Table 7**

**Nuchal Translucency (NT)**

<b>Nuchal Translucency</b>	<b>Number</b>	<b>%</b>
< 95 <sup>th</sup> centile	48	96
> 95 <sup>th</sup> centile	2	4

**Fig 31: Nuchal Translucency (NT)**



96% of pregnant women had NT less than 95<sup>th</sup> centile.

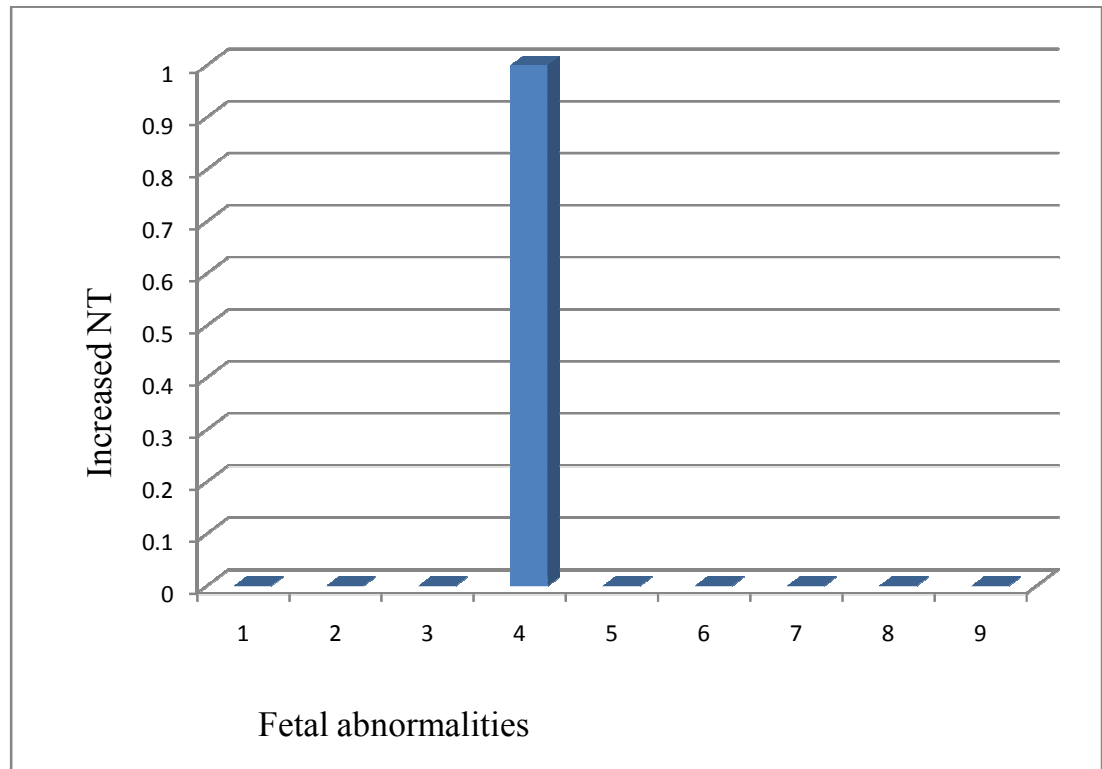
4% of pregnant women had NT more than 95<sup>th</sup> centile.

**Table 8**

**Association of increased NT (> 95th centile) with fetal abnormalities**

<b>Fetal abnormalities</b>	<b>Increased NT (n=2)</b>
1.Trisomy 21	-
2. Trisomy 18	-
3.Trisomy 13	-
4.Cardiac defects	1(50%)
5.Pulmonary defects	-
6.Abdminal wall defects	-
7.Skeletal defects*	-
8.Genetic syndromes	-
9. Fetal loss	-

**Fig 32: Association of increased NT (> 95th centile) with fetal abnormalities**



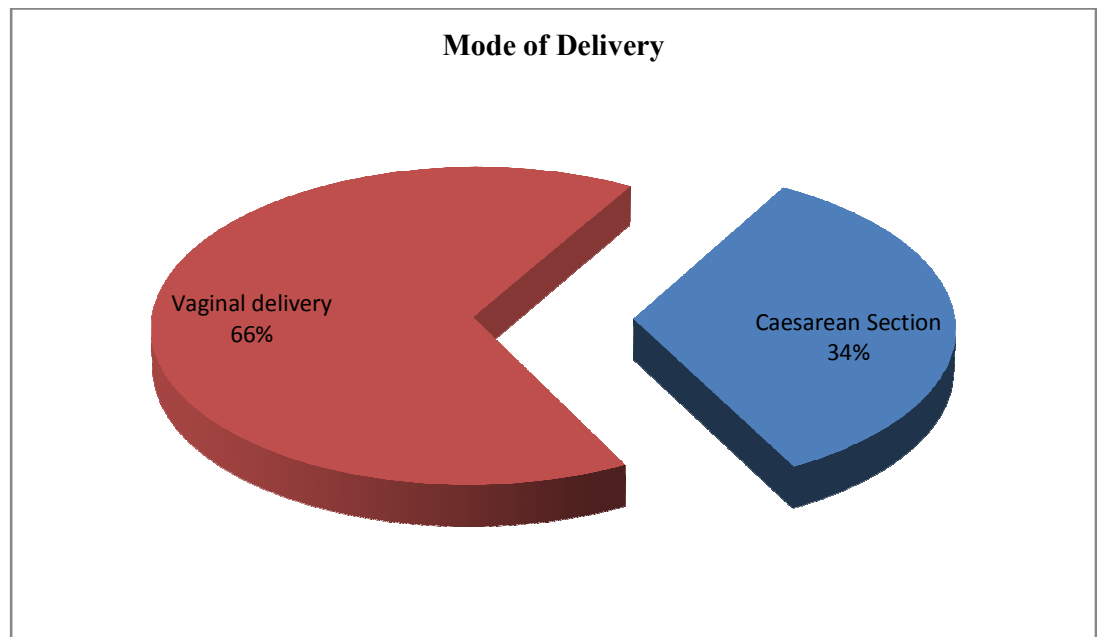
Out of the two mothers with increased NT one of the baby had cardiac defect(50%), other baby was normal.

**Table 9**

**Mode of delivery**

<b>Mode</b>	<b>Number</b>	<b>%</b>
Caesarean Section	17	34
Vaginal delivery	33	66

**Fig 33: Mode of delivery**



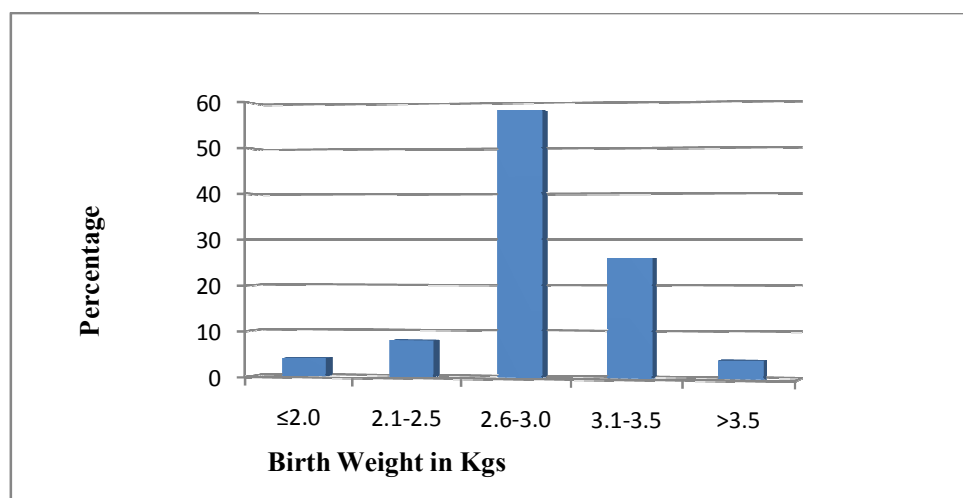
66% of pregnant women delivered vaginally and Cesarean section was done in 34%.

**Table 10**

**Birth weight**

Birth weight in kgs	Number	%
$\leq 2.0$	2	4
2.1-2.5	4	8
2.6-3.0	29	58
3.1-3.5	13	26
$> 3.5$	2	4

**Fig 34: Birth weight**



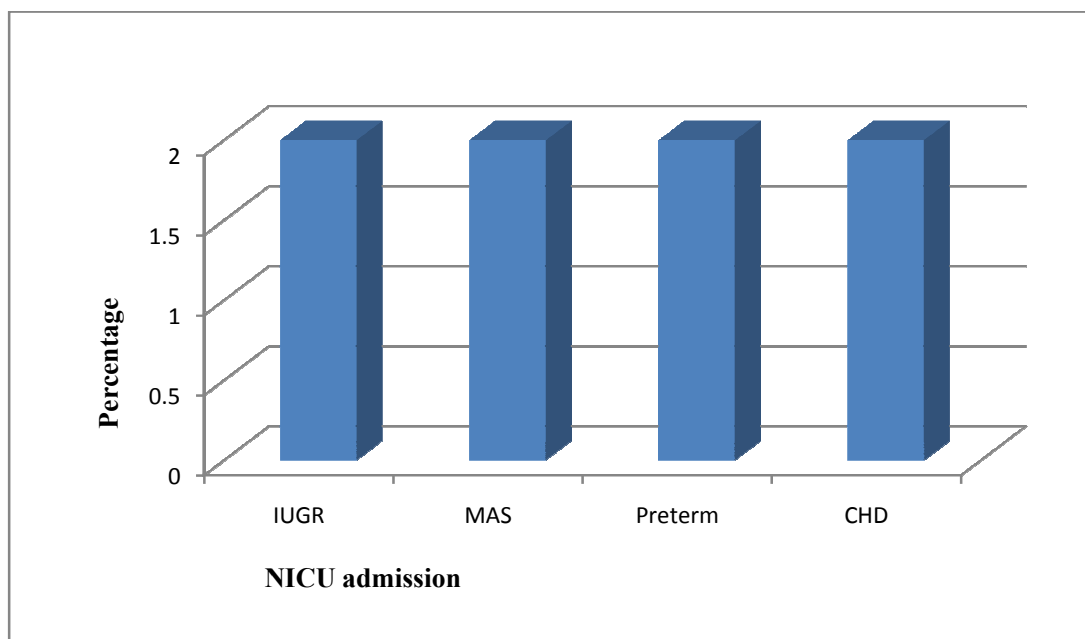
Majority of babies had birth weight between 2.6-3 kgs (58%), 26% between 3.1-3.5 kgs, 8% between 2.1-2.5 kgs, 4% weighed  $> 3.5$  kgs and 4% weighed  $\leq 2$  kgs.

**Table 11**

**NICU Admission**

NICU admission	Number	%
IUGR	1	2
MAS	1	2
Preterm	1	2
CHD	1	2

**Fig 35: NICU Admission**



8% of babies needed NICU admission out of which 2% were due to MAS, 2% were due to preterm, 2% were due to IUGR and 2% were due to Congenital heart disease (CHD).

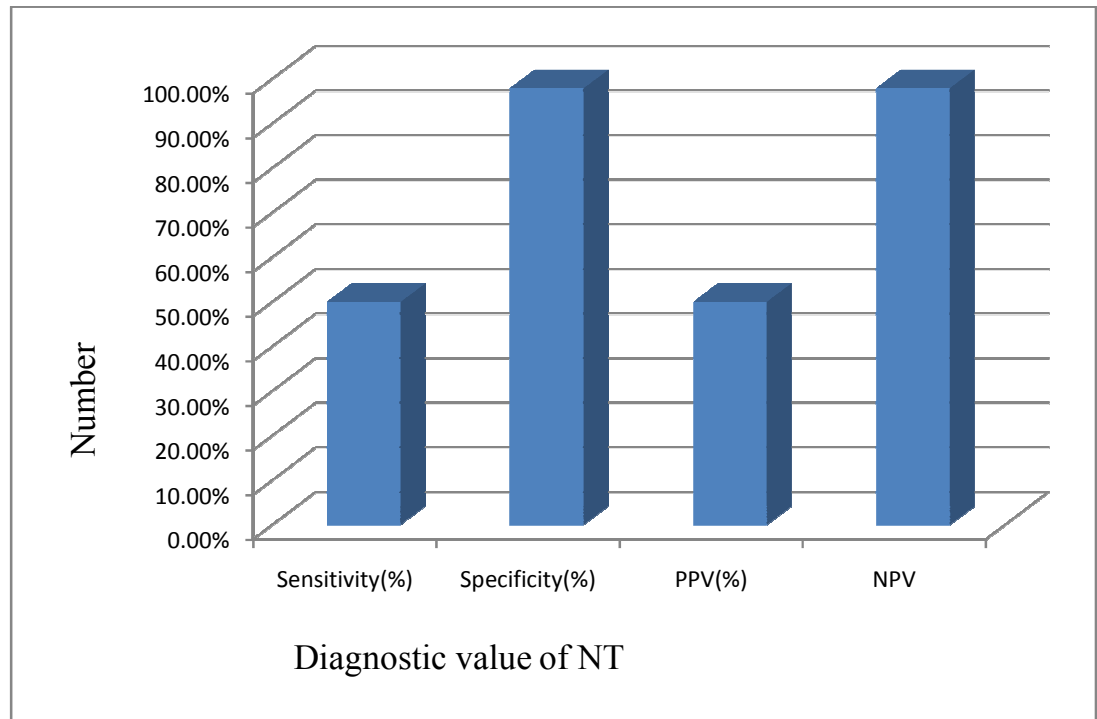


**Table 12**

**Diagnostic value of increased NT in relation to fetal abnormalities**

<b>Diagnostic value of NT</b>	<b>Number</b>
True Positives	1
True Negatives	47
False Negative	1
False Positive	1
Sensitivity(%)	50.00%
Specificity(%)	97.92%
PPV(%)	50.00%
NPV	97.92%
p value	0.017

**Fig 36: Diagnostic value of increased NT in relation to fetal abnormalities**



Fetal abnormalities are significantly related to NT with  
 $p = 0.017$

## *DISCUSSION*

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## **DISCUSSION**

Antenatal screening of nuchal translucency at 10-14 weeks of gestation is done for identification of fetal anomalies at the earliest.

There are various methods of screening for fetal anomaly such as I trimester scan, II trimester anomaly scan, fetal echocardiography, maternal serum markers and invasive testing like cordocentesis, celocentesis, CVS, amniocentesis.

The ideal test for all pregnant mothers should be non-invasive, less expensive and should give reliable information at the earliest regarding the fetal anomaly, so that the affected women would be offered an early option of termination of pregnancy. The screening test should not affect the fetus and the pregnant mothers and should not cause fetal loss. Invasive methods are associated with fetal loss and fetal limb reduction defects which causes mental trauma to the pregnant mothers. All the above conditions of an ideal screening test is fulfilled by Nuchal Translucency.

Nuchal translucency increases with increasing gestational age. It is measured by ultrasonogram and interference is made correspondingly. Any value of nuchal translucency  $>95^{\text{th}}$  centile for the gestational age is increased nuchal translucency.

In this study majority of the women were between 21-25 years(50%), 34% were between 26-30 years. 56% were primigravida and 44% were multigravida. Most of the women were screened between 12-13 weeks(52%), 30% were screened between 13-14 weeks, 18% were screened between 11-12 weeks.

22% of pregnant women had significant past obstetric complications like I and II trimester loss, IUD, preterm delivery, IUGR, GDM, GHT, anomalous baby. Rest of the pregnant women had no previous obstetric complication.

48 pregnant women in our study(96%) had normal NT valve( $<95^{\text{th}}$  centile). 2 of the pregnant women in our study(4%) had increased NT valve( $>95^{\text{th}}$  centile). All the women under study were followed with Anamoly scan and fetal echocardiography.

Among the 2 pregnant women with increased NT, one of the baby had isolated cardiac defect(ASD) and the pregnancy was continued. Other baby had no anomaly in the follow-up scan and the pregnancy was continued. This contributes to false positive.

Among the 48 pregnant women with normal NT, one of the baby had Patent ductus arteriosus. Rest of babies were normal. Increased NT identifies the group at risk not only for chromosomal abnormalities but also for major cardiac defects. Our study needed a specialist for NT measurement and fetal echocardiography. Now a days there are advanced ultrasound machines in which detailed cardiac scanning is possible even in I trimester of pregnancy.

In our study 50% of major cardiac abnormalities are associated with increased NT measured at 10-14weeks of gestation. No fetal aneuploidy was detected in our study, as the study population was only 50 and the incidence of Down syndrome is 1 in 600-700.

In spite of NT, I trimester ultrasonogram is also used to confirm fetal viability, accurate dating of pregnancy, early diagnosis of multiple pregnancy and detection of major structural anomalies, condition of uterus, adnexa and cervical length.

NT measurement done in I trimester was traditionally used for fetal aneuploidy but also identifies major cardiac defects, skeletal defects and other structural defects. The decision for termination can be made from the non-invasive testing itself. If NT is increased, other sonographic markers of screening in I trimester along with maternal serum markers can be done. If still patient is in high risk group, termination can be opted. NT measurement combined other sonographic markers would be ideal consideration for I trimester termination.

*CONCLUSION*

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## **CONCLUSION**

1. NT measurement is a non invasive, reliable screening tool available in the I trimester to determine the fetus at risk for fetal aneuploidy, cardiac defects, structural defects, genetic syndromes.
2. NT measurements in I trimester has pushed the prenatal screening for fetal anomalies from II trimester to the I trimester.
3. Apart from Nuchal Translucency, I trimester ultrasonogram is also used to confirm fetal viability, accurate dating of pregnancy, early diagnosis of multiple pregnancy and detection of major structural anomalies, condition of uterus, adnexa and cervical length.
4. Normal fetal NT in I trimester reassures the pregnant women that her fetus is not at high risk for fetal anomalies.
5. Measurement of normal NT reduces the number of invasive procedures like CVS, amniocentesis and cordocentesis
6. Measurement of increased NT provides women with affected fetuses an early termination option.

7. Increased Nuchal Translucency identifies the women at risk group and should be followed up carefully with other screening modalities.
8. The number of pregnant women were only 50. Study should continue with large number of patients to formulate a definitive protocol.

## *SUMMARY*

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## SUMMARY

It is a prospective clinical study, 50 pregnant women were selected randomly between 10-14weeks of gestation over a period of one year, attending the antenatal out-patient department of obstetrics and gynaecology at Coimbatore Medical College Hospital.

Screening for fetal NT was performed for all the 50 pregnant women between 10-14weeks of gestation. NT>95<sup>th</sup> centile are considered as screen positive. They are considered to be at high risk for fetal anomalies. All the women with normal or increased NT were followed up with anomaly scan at 20-22weeks of gestation and with 2-D fetal echocardiography at 22-24weeks of gestation.

The outcome of increased NT was predicted with anomaly scan and fetal echocardiography. Statistical analysis was carried out to test the significance of fetal anomalies in relation to increased NT and to find the diagnostic value.

In our study 50% of women were between 21-25 years, 34% were between 26-30 years, 8% were <20 years, 6% were between 31-35 years and 2% were >35 years.

Although the risk of trisomy 21 is increased after the age of 35, 80% of down's babies were born to women under the age of 35. So all women in reproductive age group are included in this study. The screen positive value of NT is >95<sup>th</sup> centile which was present in this study between the age of 26-30 years.

In this study 56% were primigravida, 44% were multigravida.

In this study NT scan were performed mostly at the gestational age of 12.1-13 weeks(52%), 30% done at 13.1-14 weeks of gestation, 18% at 11-12 weeks of gestation.

Any significant risk factor in the past pregnancy was considered and 22% had significant obstetric illness in previous pregnancies.

48 pregnant women in our study(96%) had normal NT value(<95<sup>th</sup> centile). 2 of the pregnant women in our study(4%) had increased NT value(>95<sup>th</sup> centile). Among the 2 pregnant women with increased NT, one of the baby had isolated cardiac defect(ASD) and the pregnancy was continued. Other baby had no anomaly in the follow-up scan and the pregnancy was continued. This contributes to false positive.

Among the 48 pregnant women with normal NT, one of the baby had Patent ductus arteriosus. Rest of babies were normal.

The sensitivity of fetal nuchal translucency >95<sup>th</sup> centile used for screening anomalies is 50%, specificity is 97.92%, positive predictive value is 50%, negative predictive value is 97.92% and the p value is 0.017. Fetal anomalies are significantly related to increased value of nuchal translucency.

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## *ANNEXURES*

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## PROFORMA

Name:

Age:

Husband's Name:

Address:

O. P. No:

Education:

Occupation:

Monthly Income:

### PRESENTING COMPLAINTS:

OBH: ML Consanguinous/Non- Consanguinous G P L A

Sl. No.	Year	Pregnancy outcome	Delivery outcome	Baby outcome

H/O Previous Unexplained Fetal/Neonatal Death

H/o Congenital Anomalies In The Previous Child

MH: AOM

CYCLES-REGULAR

### **PAST HISTORY**

Any H/o Congenital defects in the mother

H/o DM /HTN / TB/ Epilepsy /Asthma/ Prev. surgery / Allergy to drug

### **FAMILY HISTORY**

H/o any congenital anomalies / Mental Retardation in both the families.

H/o Unexplained Fetal / Neonatal Death/ Recurrent Pregnancy Loss

### **GENERAL PHYSICAL EXAMINATION**

Pallor / Icterus / Edema / Clubbing / Cyanosis / Lymphadenopathy

PR                      BP

Ht                      Wt

Breast                      Thyroid

### **SYSTEMIC EXAMINATION**

CVS

RS

CNS

### **OBSTETRIC EXAMINATION**

Sl. No.	Date	WT	BP	P/A	P/V
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## INVESTIGATIONS

### A. Routine:

Hb%	HIV	BT
PCV	HbsAg	CT
Urine Routine	VDRL	
Blood Group and RH Typing	RBS	

### B. ULTRASOUND

#### 1. 10-14 weeks scan

Gestation Sac	Gestation Age
CRL	Both Adnexa
Anomalies	NT

#### 2. IF NT INCREASED OR NORMAL → II TRIMESTER SCAN AND FETAL ECHOCARDIOGRAPHY DONE

#### 3. II TRIMESTER SCAN:

BPD	GESTATION AGE
HC	FHR
AC	PLACENTA
FL	EFW
EDD	

#### CONGENITAL ANOMALIES

#### **4. FETAL ECHO**

IF IN II TRIMESTER SCAN

- NO CONGENITAL ANOMALIES DETECTED → CONTINUATION OF PREGNANCY
- LETHAL CONGENITAL ANOMALIES → TERMINATION

#### **5.III TRIMESTER SCAN:**

BPD

EFW

FL

PLACENTA

HC

FHR

AC

AFI

GESTATION AGE

PRESENTATION

EDD

**Nature of delivery**

INDUCTION      \* INDICATION

\* METHOD

SPONTANEOUS

**MODE OF DELIVERY:**

- VAGINAL
- ABDOMINAL

**UMBILICAL CORD / PLACENTA**

**EVALUATION OF BABY BY PAEDIATRICIAN:**

SEX

WT

DOB

TOB

APGAR

ANY CONGENITAL ANOMALIES / CARDIAC DEFECTS

PERINATAL MORTALITY

## CONSENT FORM

Here by I volunteer and to participate in this study  
“**A STUDY OF NUCHAL TRANSLUCENCY IN  
EARLY PREGNANCY**” was fully explained about the  
nature of this study by the doctor, knowing which I  
Mr/Mrs\_\_\_\_\_ fully consent to  
volunteer in this study.

Date:

Place:

Signature of the Volunteer



## ஒப்புதல் படிவம்

பெயர் :

பாலினம் :

வயது :

முகவரி :

அரசு கோவை மருத்துவக் கல்லூரியில் மகப்பேறு மருத்துவ துறையில் பட்ட பயிலும் மாணவி அவர்கள் மேற்கொள்ளும் "பிறக்கப் போகும் குழந்தைக்கு பிறவிக் குறைபாடு உள்ளதா என்பதை அறியும் நியூக்கள் டிரேன்ஸ்லுசன்சீ என்னும் ஸ்கேன் அளவீடு" குறித்த ஆய்வில் செய்முறை மற்றும் அனைத்து விவரங்களையும் கேட்டுக் கொண்டு எனது சந்தேகங்களை தெளிவுப்படுத்திக் கொண்டேன் என்பதை தெரிவித்துக் கொள்கிறேன்.

நான் இந்த ஆய்வில் முழு சம்மதத்துடன், சுய சிந்தனையுடனும் கலந்து கொள்ள சம்மதிக்கிறேன்.

இந்த ஆய்வில் என்னுடைய அனைத்து விபரங்கள் பாதுகாக்கப்படுவதுடன் இதன் முடிவுகள் ஆய்விதழில் வெளியிடப்படுவதில் ஆட்சேபனை இல்லை என்பதை தெரிவித்துக் கொள்கிறேன். எந்த நேரத்தில் இந்த ஆய்விலிருந்து நான் விலகிக் கொள்ள எனக்கு உரிமை உண்டு என்பதையும் அறிவேன்.

இடம் :

கையொப்பம்

நாள் :

Sl. No	Name	Age	IP No	Parity	Gestational Age at NT	NT Value in mm	Anomaly Scan at 20-22 wks	Fetal Echo at 22-24 wks	Risk factor present at NT Scan	Risk factor developed later	Medical history	Significant past obstetric history	Anomaly in fetal mily	Previous Anomaly	Anomaly present now	Outcome & mode of delivery	Baby Details			
																	Sex	Birth Wt in Kg	Apgr	Complications
1	Kohila	19	47810	Primi	12w 3d	0.89	Normal	Normal	Nil	Nil	Nil	-	-	-	-	LN	Me h	2.7	8/10	Nil
2	Divya	20	47719	Primi	11w 6d	1.4	Normal	Normal	Nil	Nil	Nil	-	-	-	-	LN	Fch	2.7	8/10	Nil
3	Kanimo zhi	33	46801	G <sub>3</sub> P <sub>1</sub> L <sub>1</sub> A <sub>1</sub>	13w 1d	1.6	Normal	Normal	Overt DM	Polyhydramnios	DM	Itrimester abortion GDM	-	-	-	LSCS	Me h	3.4	8/10	Nil
4	Palania mmal	25	47623	Primi	12w 4d	1.8	Normal	Normal	Nil	Nil	Nil	-	-	-	-	LSCS	Fch	2.7	8/10	Nil
5	Kavita	22	48210	G <sub>2</sub> P <sub>1</sub> L <sub>1</sub>	12w 3d	1.3	Normal	Normal	Nil	Nil	Nil	Prev. Preterm	-	-	-	LN	Fch	2.75	8/10	Nil
6	Jaya	25	47312	G <sub>3</sub> P <sub>2</sub> L <sub>2</sub>	12w 4d	1.1	Normal	Normal	Nil	Post dated	Nil	-	-	-	-	AVD	Me h	3.15	7/10	MAS
7	Shanti	23	48001	Primi	12w 3d	1.3	Normal	Normal	Nil	GHT	Nil	-	-	-	-	LN	Fch	2.4	8/10	Nil
8	Anisha Praveen	27	48480	G <sub>3</sub> P <sub>2</sub> L <sub>2</sub>	13w4d	1.2	Normal	Normal	Nil	Recurrent GHT	Nil	GHT	-	-	-	LN	Me h	3.6	8/10	Nil
9	Sasikala	31	47693	Primi	12w 4d	1.3	Normal	Normal	Nil	Postdated	Nil	-	-	-	-	LSCS	Me h	2.8	8/10	Nil
10	Dhanalaksmi	29	33578	G <sub>2</sub> P <sub>1</sub> L <sub>1</sub>	12w 3d	1.7	Normal	Normal	Perv. LSCS	Nil	Nil	-	-	-	-	LSCS	Fch	2.75	8/10	Nil

Sl. No	Name	Age	IP No	Parity	Gestational Age at NT	NT Value in mm	Anamoly Scan at 20-22 wks	Fetal Echo at 22-24 wks	Risk factor present at NT Scan	Risk factor developed later	Medical history	Significant obstetric history	Anamoly in family	Previous Abnormal	Anamoly present now	Outcome of delivery	Baby Details			
																	Sex	Birth weight	Apgar	Complications
11	Rekha	21	47133	Primi	12w 2d	1.8	Normal	Normal	Nil	Oligohydramnios	Nil	-	-	-	-	LN	Female	2.75	8/10	Nil
12	Rajeshwari	25	47671	G <sub>2</sub> P <sub>1</sub> L <sub>0</sub>	11w 5d	1.4	Normal	Normal	Nil	Posdated	Nil	IUD at 36wks	-	-	-	LN	Male	2.7	8/10	Nil
13	Muneshwari	27	46951	Primi	13w 3d	4.9	Normal	ASD	Nil	Nil	Nil	-	-	-	-	LN	Female	2.85	8/10	CHD-ASD
14	Elavarasi	24	47902	G <sub>2</sub> P <sub>2</sub> L <sub>1</sub>	11w 4d	0.88	Normal	Normal	Prev. LSCS	GHT	Nil	-	-	-	-	LSCS	Male	2.65	8/10	Nil
15	Latha	22	48114	Primi	13w 1d	1.2	Normal	Normal	Nil	Preterm Labour	Nil	-	-	-	-	LN	Male	2.3	7/10	LBW
16	Parvathi	33	46991	Primi	13w 3d	1.05	Normal	Normal	Nil	Nil	Nil	-	-	-	-	LN	Female	2.6	7/10	Nil
17	Kavitha	29	48721	G <sub>3</sub> P <sub>1</sub> L <sub>1</sub> A <sub>1</sub>	12w 3d	1.1	Normal	Normal	Nil	Posdated	Nil	Spontaneous abortion at 10wks	-	-	-	LSCS	Female	3.6	8/10	Nil
18	Madhumalar	26	47080	Primi	12w 1d	1.1	Normal	Normal	Nil	GHT	Nil	-	-	-	-	LSCS	Female	2.7	7/10	MAS
19	Chellamani	25	48001	G <sub>2</sub> P <sub>2</sub> L <sub>1</sub>	13w 5d	1.6	Normal	Normal	Prev. LSCS	Oligohydramnios	Nil	-	-	-	-	LSCS	Female	2.6	8/10	Nil
20	Indrani	25	47584	Primi	11w 4d	1.0	Normal	Normal	Nil	Nil	Nil	-	-	-	-	AVD	Male	3.3	8/10	Nil

Sl. No	Name	Age	IP No	Parity	Gestational Age at NT	NT Value in mm	Anomaly Scan at 20-22 wks	Fetal Echo at 22-24 wks	Risk factor present at NT Scan	Risk factor developed later	Medical history	Significant obstetric history	Anomaly in family	Previous Abnormal	Anomaly present now	Outcome & mode of delivery	Baby Details			
																	Sex	Birth Wt in kg	Appar	Complications
21	Vasantha	26	47565	G <sub>3</sub> P <sub>1</sub> L <sub>1</sub> A <sub>1</sub>	13w 4d	1.3	Normal	Normal	Prev. LSCS	Nil	Nil	1 <sup>st</sup> trimester Abortion	-	-	-	LSCS	Fch	2.9	8/10	Nil
22	Fathima	19	46877	Pri mi	12w 58	1.5	Normal	Normal	Nil	Nil	Nil	-	-	-	-	LSCS	Fch	2.6	8/10	Nil
23	Poomima	30	47254	Pri mi	13w 1d	1.1	Normal	Normal	Nil	Nil	Nil	-	-	-	-	LN	Fch	2.7	8/10	Nil
24	Aruna	23	47465	Pri mi	12w 5d	1.3	Normal	Normal	Nil	Nil	Nil	-	-	-	-	LN	Meh	2.8	8/10	Nil
25	Ramya	28	48102	G <sub>2</sub> P <sub>1</sub> L <sub>1</sub>	12w 5d	4.1	Normal	Normal	Prev. LSCS	Nil	Nil	-	-	-	-	VBA C	Meh	2.85	7/10	Nil
26	Deepa	21	46891	Pri mi	13w 6d	2.1	Normal	Normal	Nil	GHT	Nil	-	-	-	-	LN	Fch	2.7	8/10	Nil
27	Pechiammal	25	47786	G <sub>2</sub> P <sub>1</sub> L <sub>1</sub>	13w 5d	1.2	Normal	Normal	Prev. LSCS	Nil	Nil	-	-	-	-	LSCS	Meh	3.4	8/10	Nil
28	Kunjammal	29	48442	Pri mi	12w 3d	0.9	Normal	Normal	Nil	GD M	Nil	-	-	-	-	LSCS	Fch	3.3	8/10	Nil
29	Geetha	26	48956	G <sub>3</sub> P <sub>1</sub> L <sub>1</sub> A <sub>1</sub>	13w 2d	1.3	Normal	Normal	Nil	IUG R	Nil	1 <sup>st</sup> trimester abortion	-	-	-	LN	Meh	1.85	6/10	IUGR
30	Abirami	22	47553	G <sub>2</sub> P <sub>1</sub> L <sub>1</sub>	13w 5d	1.85	Normal	Normal	Nil	PRO M	Nil	-	-	-	-	LN	Fch	2.8	8/10	Nil

Sl. No	Name	Age	IP No	Parity	Gestational Age at NT	NT Value in mm	Anamoly Scan at 20-22 wks	Fetal Echo at 22-24 wks	Risk factor present at NT Scan	Risk factor developed later	Medical history	Significant past obstetric history	Anamoly in family	Previous Anamoly	Anamoly present now	Outcome & mode of delivery	Baby Details			
																	Sex	Birth Wt in kg	Apgar	Complications
31	Thavamani	28	47664	Primi	13w 5d	1.3	Normal	Normal	Nil	Nil	Nil	-	-	-	-	LN	Fch	3.2	8/10	Nil
32	Bannari	24	48325	G <sub>3</sub> P <sub>1</sub> L <sub>1</sub> A <sub>1</sub>	12w 3d	1.85	Normal	Normal	Prev. LSCS	Nil	Nil	I trimester abortion	-	-	-	LSCS	Fch	2.9	8/10	CHD-PDA
33	Kalavathi	27	47341	Primi	11w 5d	1.0	Normal	Normal	Nil	Nil	Nil	-	-	-	-	LN	Mch	2.9	7/10	Nil
34	Nalini	36	46861	Primi	13w 5d	1.3	Normal	Normal	Elderly Primi	Nil	Nil	-	-	-	-	LSCS	Mch	2.7	8/10	Nil
35	Nithys	23	47529	G <sub>3</sub> P <sub>1</sub> L <sub>1</sub> A <sub>1</sub>	12w 3d	1.1	Normal	Normal	Nil	Nil	Nil	Prev. anencephaly terminated	-	Anencephaly	-	LN	Mch	2.6	8/10	Nil
36	Pavithara	18	47281	Primi	12w 2d	1.1	Normal	Normal	Nil	Nil	Nil	-	-	-	-	LN	Mch	2.65	8/10	Nil
37	Priyadharshini	24	46894	G <sub>2</sub> P <sub>1</sub> L <sub>1</sub>	12w 5d	1.1	Normal	Normal	Nil	Nil	Nil	-	-	-	-	LN	Fch	3.25	8/10	Nil
38	Sajitha	23	47740	Primi	13w 3d	1.1	Normal	Normal	Nil	Oligo hydramnios	Nil	-	-	-	-	LSCS	Fch	2.4	8/10	Nil
39	Durga	24	47608	G <sub>2</sub> P <sub>1</sub> L <sub>1</sub>	12w 4d	1.2	Normal	Normal	Nil	Postdated	Nil	-	-	-	-	LN	Fch	3.1	8/10	Nil
40	Maragathavalli	26	48732	Primi	12w 3d	1.3	Normal	Normal	Nil	GHT	Nil	-	-	-	-	LN	Mch	3.25	8/10	Nil

Sl. No	Name	Age	IP No	Parity	Gestational Age at NT	NT Value in mm	Ana moly Scan at 20-22 wks	Fetal Echo at 22-24 wks	Risk factor present at NT Scan	Risk factor developed later	Medical history	Significant obstetric history	Ana moly in family	Previous Ana moly	Ana moly present now	Outcome & follow up	Baby Details			
																	Sex	Bir th Wt in	Ag ar	Co mpla cat ion
41	Mangayarkarasi	22	47613	Primi	12w 3d	1.1	Normal	Normal	Hypothyroidism	Nil	Nil	-	-	-	-	LN	Mch	2.7	8/10	Nil
42	Nambikaimary	23	48001	Primi	11w 3d	0.96	Normal	Normal	Nil	PROM	Nil	-	-	-	-	LN	Mch	2.6	8/10	Nil
43	Selvi	24	47406	Primi	11w 6d	1.1	Normal	Normal	Nil	Nil	Nil	-	CHD	-	-	LS CS	Mch	3.25	8/10	Nil
44	Rubiyabanu	24	46903	Primi	12w 5d	0.95	Normal	Normal	Nil	PROM	Nil	-	-	-	-	LN	Mch	3.1	8/10	Nil
45	Nisha	24	47671	Primi	11w 3d	1.05	Normal	Normal	Nil	PROM	Nil	-	-	-	-	LN	Mch	2.3	8/10	Nil
46	Habebunisha	23	47814	G <sub>2</sub> P <sub>1</sub> L <sub>1</sub>	12w 1d	1.3	Normal	Normal	Nil	Nil	Nil	Prev GHT IUGR	-	-	-	LN	Mch	2.7	8/10	Nil
47	Thulasi	26	48121	Primi	12w 5d	1.15	Normal	Normal	Nil	PPROM	Nil	-	-	-	-	LN	Mch	1.5	8/10	PT B
48	Sangeetha	28	47551	G <sub>2</sub> P <sub>1</sub> L <sub>1</sub>	11w 4d	0.8	Normal	Normal	Prev. LSCS	Breech	Nil	-	-	-	-	LS CS	Fch	2.75	8/10	Nil
49	Mugammal	27	48449	G <sub>2</sub> P <sub>1</sub> L <sub>1</sub>	12w 5d	1.1	Normal	Normal	Nil	Nil	Nil	-	-	-	-	LN	Fch	3	8/10	Nil
50	Anadhi	28	47905	G <sub>2</sub> P <sub>1</sub> L <sub>1</sub>	12w 3d	0.85	Normal	Normal	Nil	Nil	Nil	-	-	-	-	LN	Mch	3.25	8/10	Nil

## KEY TO MASTER CHART

G	→	Gravida
P	→	Parity
L	→	Live child
A	→	Abortion
DM	→	Diabetes mellitus
GHT	→	Gestational Hypertension
Prev	→	Previous
LSCS	→	Lower segment cesarean section
PROM	→	Prelabour rupture of membranes
PPROM	→	Preterm prelabour rupture of membranes
LN	→	Labour Natural
AVD	→	Assisted Vaginal Delivery
VBAC	→	Vaginal Birth After Cesarean
PTB	→	Preterm Birth
IUD	→	Intrauterine Death
IUGR	→	Intra uterine growth restriction
MAS	→	Meconium Aspiration syndrome
LBW	→	Low birth weight
CHD	→	Congenital heart disease
ASD	→	Atrial septal defect
PDA	→	Patent ductus arteriosus